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**Information for Users [PRE-4]**

# The Service

The Laboratory Genetics department forms part of the West of Scotland Centre for Genomic Medicine and provides a comprehensive diagnostic genetics service for the patients of the West of Scotland (population >2.7 million) and specialised testing for particular disorders to the whole of Scotland, the UK and overseas. The West of Scotland Centre for Genomic Medicine also includes the Clinical Genetics Service, which is co-located.

The laboratory is situated on Level 2 of the Laboratory Medicine Building at the Queen Elizabeth University Hospital, Glasgow and encompasses cytogenetics and molecular diagnostic testing for the specialist diagnosis and/ or monitoring of patients with constitutional (prenatal and postnatal) and acquired (malignant) genetic abnormalities in hereditary genetic disease, solid tumours, and adult and childhood leukaemia.

The laboratory is a member of the Scottish Strategic Network for Genomic Medicine, with laboratories also located in Aberdeen, Dundee, and Edinburgh. The laboratories in each of these four centres are funded by National Services Division of NHS Scotland and are commissioned to work together to provide a comprehensive genetics service to the patients of Scotland. In addition, the laboratory sources genetic testing from other laboratories when required ensuring our patients have access to specialist genetic tests from other UK accredited laboratories, where appropriate.

The Laboratory Genetics department is an accredited laboratory. The scope of services offered for laboratory testing is accredited by the United Kingdom Accreditation Service (UKAS) to ISO 15189 standards (Medical Laboratory 8290).

**Information for Users**

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# Contacting the laboratory

**Postal address for correspondence and samples**

West of Scotland Centre for Genomic Medicine

Laboratory Genetics

Level 2B

Laboratory Medicine

Queen Elizabeth University Hospital

Glasgow

G51 4TF

**Website address**

[www.nhsggc.scot/laboratory-genetics](http://www.nhsggc.scot/laboratory-genetics)

**Other ways of contacting the laboratory**

Tel: 0141 354 9300

Email: Genetic.Laboratories@ggc.scot.nhs.uk

The department’s email account is monitored daily and is suitable for the receipt of patient-identifiable information. Personal or identifiable information **should not** be added to the subject line when emailing. Patient identifiable information should only be sent to the laboratory from secure accounts such as scot.nhs.uk, nhs.scot or nhs.net accounts. Do NOT send patient identifiable information to the laboratory from any other email provider.

**Voicemail**

When diverted to voicemail, please leave a message and your contact telephone number. Someone from the department will get back to you as soon as possible.

**Department working hours**

Monday to Friday 8.30 a.m. – 5.00 p.m.

The laboratory does not offer an out of hours service, but it may be possible to arrange the analysis of urgent samples out with these times, by prior arrangement.

The department is closed during public holidays.

# Laboratory Staff

**Please refer to the website for main laboratory contacts.**

# Complaints

Should you have any comments, suggestions, cause for concern or complaints about the service you receive from the laboratory, please contact the Head of Service, Deputy Head of Service, Compliance and Resource Programme Manager or Quality Manager, using the contact details found on the website.

# Sending a sample to the laboratory

**Reasons for referral**

The laboratory uses various analysis techniques to carry out tests for a wide range of hereditary and acquired genetic disorders (haematological malignancy and solid tumours).

The types of investigation include:

* Confirmation/exclusion of a diagnosis for hereditary and acquired disorders.
* Carrier testing and risk assessment in families with a known genetic disorder.
* Presymptomatic/predictive testing in individuals at risk of a late-onset hereditary genetic disorder.
* Prenatal diagnosis of hereditary genetic conditions where appropriate.
* Pharmacogenetic testing to determine drug treatment options.
* Diagnostic tests to aid diagnosis and classification of malignant diseases e.g. lymphoma, sarcoma.
* Follow up testing to monitor treatment response or disease progression in haematological malignancy, e.g. post-transplant chimerism, minimal residual disease

The laboratory offers testing for a range of ‘core’ disorders plus a set of more specialist services for which samples are received on a supra-regional or national basis (see the relevant Test Directory available via our website).

Where tests are not available in our laboratory, we will send DNA/cDNA or appropriate samples to other accredited UK genetic laboratories for testing for a large range of hereditary genetic disorders following appropriate approval from our Clinical Genetics Service (see below) and for selected somatic disorders (e.g., AML MRD). Please contact the laboratory for details of access and availability of tests.

Details of services not currently available in the UK can be found on the web site [www.orpha.net](http://www.orpha.net) or by contacting the laboratory.

Each request accepted by the laboratory for testing is considered an agreement between the user and laboratory. Terms and Conditions of Service are available to read on our website (see above).

**Gate-keeping and funding of tests**

Access to many tests provided by other laboratories is restricted to specific referring specialities or clinicians. Clinicians may have to provide specific referral criteria and complete patient information forms, before these tests can be requested. Please contact the laboratory for details.

Please note that specimens referred out-with Scotland for testing will have a cost implication. Authorisation for testing for germline disorders is in consultation with our Clinical Genetics Service and for high value referrals approval is granted by the Scottish Strategic Network for Genomic Medicine clinical representatives. Following approval any associated costs will be met by National Services Division (NSD) of NHS Scotland. However, in some instances authorisation for testing may be declined and therefore any associated cost must be met by the referring clinician/ clinical service. Specimens referred out-with Scotland for somatic cancer are not funded by NSD.

**Consent**

***All genetic testing requires appropriate consent.*** The laboratory assumes that when sending a sample consent has been obtained by the referring clinician as outlined in our Genetics Test Request form. Testing and/or storage of genetic material must be discussed with the patient with a summary of clinical consent recorded in the patient’s health record. Please refer to the website for ‘Genetic Record of Discussion Consent Form’. For further information and guidance, please refer to *‘Consent and Confidentiality in genomic medicine -Guidance on the use of genetic and genomic information in the clinic’* which is available as a download from the British Society of Genetic Medicine website at [www.bsgm.org.uk](http://www.bsgm.org.uk).

Products of conception requiring post-mortem and/ or sensitive disposal must be accompanied by the necessary paperwork supplied by local pathology department.

The department is committed to protecting personal information and complies with NHSGGC policies and the principles of the Data Protection Act.

**Referral information**

***Test request forms*** *(and* ***test proformas*** for certain disorders) are available on request by emailing Genetic.Laboratories@ggc.scot.nhs.uk or by calling the laboratory on 0141 354 9300.

Forms are also available for printing via the website under ‘Information for healthcare professionals’

Please see the Test Directory on our website for details of tests which will require a completed proforma.

**Electronic requests** for Germline testing are also available for users within NHSGGC through TrakCare and ICE.

Referral forms must be legible and should contain the patient’s surname, forename, date of birth/ CHI number and postcode along with the reason for referral/ analysis/ test requested and a name and address for the referring clinician for reporting purposes. Referral forms received with insufficient information may be rejected.

Where no test is currently available for a referred disorder, the specimen will be accepted for DNA/ RNA extraction and storage as appropriate. The laboratory may accept samples collected for R&D projects following prior discussion and agreement with the Head of Laboratory.

The laboratory cannot issue a report without written confirmation, all verbal requests for further testing must be followed up in writing (request form or email). Please submit a formal written request within the specified turnaround times of the test requested.

After receipt and processing of the primary sample for testing, and assuming appropriate material has been stored by the laboratory, additional testing may be requested. Please contact the laboratory to discuss (see website for appropriate contact details and link to ‘[Further Analysis Request Form](https://forms.office.com/pages/responsepage.aspx?id=veDvEDCgykuAnLXmdF5JmoIYLqZPg35IoY0jMY6qcLxUQkoyMlg2N1AwNlg0NEdSMUtWWjRaT1lZQi4u)’).

**Specimen labels**

All specimens should be clearly labelled with surname, forename and date of birth/ CHI number and must match those supplied on the test referral form. If the specimen label does not match the information that is supplied on the test referral form, the ***sample will be discarded***.

Samples received without a test referral form ***will be discarded***.

Please ensure other labels e.g., those which are added to the specimen tube by other laboratories, ***do not obscure the original specimen label*** containing the patient identifiers (name and date of birth/ CHI number) or the specimen ***may be rejected***.

**Unsuitable specimens**

Samples arriving in an unsuitable condition will not be processed and will therefore be rejected by the laboratory. Unsuitable conditions include but is not limited to, samples sent in the wrong container, samples significantly delayed in transport, clotted blood samples or blood samples in an inappropriate blood tube and broken sample tubes. In such circumstances the ***sample will be discarded,*** and a repeat sample will be requested if appropriate.

Requests for genetic testing in patients who are known to have received a recent blood transfusion should be discussed with the laboratory as testing may need to be delayed.

For patients who are known to have received an allogeneic bone marrow transplant and for whom germline testing is being considered, please contact the laboratory to discuss (peripheral blood and buccal samples are not acceptable for analysis).

**Specimens from patients with blood borne viruses (Hepatitis and HIV)**

If a sample is known or suspected to be affected with HIV, Hepatitis B or Hepatitis C, it is no longer a requirement to label the specimen container as ‘danger of infection’ or ‘high risk’.

**Specimens from patients with Creutzfeldt Jakob Disease**

Specimens from individuals with a confirmed or suspected diagnosis of Creutzfeldt Jakob Disease (CJD) must be labelled as ‘danger of infection’ or ‘high risk’, with the CJD status clearly indicated on the referral form. These specimens are not processed by the laboratory for DNA extraction or tissue culture, instead they are sent to the CJD Surveillance Unit in Edinburgh for DNA extraction prior to testing.

**Specimens from patients with other suspected Group 3 and 4 pathogenic infections**

For patients with a suspected or known TB infection, please contact the laboratory to discuss. The laboratory ***cannot*** process specimens from patients who have or are suspected as having any other Group 3 or Group 4 pathogenic infections (other than those specified above).

**Packaging and transportation**

It is the responsibility of those taking and dispatching specimens to the laboratory to ensure that these samples are sent in accordance with any national guidelines and/ or local policies for packaging, labelling and transport of biological material.

Specimens sent internally using the Greater Glasgow and Clyde specimen transport system should be placed in sealed plastic bag containing absorbent material, with the accompanying referral form placed in a separate compartment.

Blood and fluid specimens sent through the post should be packaged in accordance with PI 650 and UN3373 regulations. Specimens should be wrapped in absorbent material and then placed inside a rigid leak-proof primary receptacle, which should then be placed inside a rigid leak-proof secondary receptacle e.g., bio-bottle. The package must then be placed into outer package and should be clearly labelled with the laboratory’s address and the sender details. The outer packaging must also be clearly labelled with the words ‘BIOLOGICAL SUBSTANCE – CATEGORY B’.

All samples should be sent to the laboratory as soon as possible after being taken. If this is not possible, we would recommend storage at 4-8oC for blood specimens, and at room temperature for bone marrow, amniotic fluid, CVS and tissue specimens, until you are able to send to the laboratory. **N.B.** DNA/ RNA quality may be affected by delays in transit which may compromise testing.

# Turnaround times

Reporting times listed are based on calendar days. These range from 3 to 112 days depending on urgency and complexity of testing. The recommended turnaround times for each test is detailed in the Genomic Test Directories for Germline and Somatic referrals (please refer to the website).

***Where more urgent testing is required for treatment decisions, please contact the laboratory to discuss.***

# Reporting of Results

NHS Greater Glasgow and Clyde Laboratory Genetics predominantly releases electronic clinical reports and paper reports are only issued in rare circumstances where no electronic route is viable. Routinely, clinical reports are released via Clinical Portal/SCI Store, but reports can be released via email for non-GGC NHS Health Boards that do not have SCI-to-SCI store access or for particular clinicians where there is an exceptional reason why they cannot access Clinical Portal/SCI Store. When a genetics report is issued to the GGC Clinical Portal, an email notification can be sent to a chosen email address of the referring clinician. However, wherever possible, this email address should be an admin, departmental or generic email address that reduces the clinical risk around notifications only going to a single individual. The same applies to email addresses for referrers from non-GGC NHS Health Boards where the Reports are emailed, wherever possible individual email addresses should not be used unless the clinician assumes responsibility for the associated clinical risk. Referrals marked as urgent can be telephoned following special arrangement with the laboratory, however telephone reporting is not available routinely for urgent referrals. For queries regarding electronic reporting please contact the laboratory directly.

Acute lymphoblastic leukaemia minimal residual disease (MRD) results are reported directly to the referring clinician and their nominated clinical team.

Where a test is out with UKAS scope of accreditation, the issued report will include the following statement ***\*Please note that this test is not currently UKAS accredited and interpretations expressed herein are outside the scope of UKAS accreditation****.*

# Clinical advice and interpretation

Where advice on appropriate testing, result interpretation or clinical advice is required please contact the laboratory (see laboratory contacts listed on the laboratory website) or Clinical Genetics on 0141 354 9200.

# Quality control and accreditation

The laboratory participates in appropriate external quality assessment schemes run by GenQA (Genomics Quality Assessment), EMQN (European Molecular Genetics Quality Network) and other appropriate UK and European EQA schemes to cover the scope of testing. Details of performance in these EQA schemes can be obtained by contacting the Quality Manager. Where an Accredited EQA scheme or formal inter laboratory comparison (ILC) programme is not available (e.g. for a rare disorder, or a new technique) the laboratory chooses an alternative approach to provide objective evidence for determining the acceptability of examination results. The laboratory also conforms to the best practice guidelines issued by EMQN and the Association for Clinical Genomic Science (ACGS), which is a constituent organisation of the British Society of Genetic Medicine (BSGM).

The scope of services offered for laboratory testing is accredited by the United Kingdom Accreditation Service (UKAS) to ISO 15189 standards (Medical Laboratory 8290). Details of the scope are available via the UKAS website [www.ukas.com](http://www.ukas.com). The UKAS symbol appears on report templates and where tests are out with scope and therefore not accredited, this is clearly detailed on the relevant report.

# Specimen requirements for germline (constitutional) investigations

**Blood samples for postnatal germline (constitutional) disorders**

Blood for molecular DNA investigations should be collected in potassium EDTA (KE) tubes. Blood for cytogenetics chromosome investigation should be collected in Lithium Heparin tubes. If requesting both molecular DNA and cytogenetic investigations, TWO blood specimens are required; one Lithium Heparin tube and one EDTA (KE) tube.

|  |  |
| --- | --- |
| **EDTA (KE) specimen tube** | **Volume of whole blood** |
| Adult | 3ml |
| Paediatric | 3ml |
| Newborn | 1ml |

|  |  |
| --- | --- |
| **Lithium Heparin specimen tube** | **Volume of whole blood** |
| Adult | 3ml |
| Paediatric | 3ml |
| Newborn | 1ml |

**Blood samples for postnatal germline (constitutional) microarray analysis**

For microarray analysis, send one EDTA (KE) tube as shown in the table below. If the patient is 6 months or younger or requires sedation/general anaesthetic for blood collection, please also send a lithium heparin sample of similar volume. This will be stored in case it is required for cytogenetic/FISH investigations following microarray findings.

|  |  |
| --- | --- |
| **Patient** | **Volume of whole blood in EDTA (KE)** |
| Adult | 3ml  |
| Paediatric | 3ml plus Lithium Heparin specimen tube **\***  |
| Newborn | 1ml plus Lithum Heparin specimen tube  |

*\** **If** $\leq $ **6 months old and/or requires sedation/general anaesthetic for blood collection**

*Note: Please contact the laboratory if the patient has previously had a bone marrow transplant as this could affect the results of germline genetic testing.*

**DNA from other laboratories for hereditary genetic disorders**

A minimum of 2µg of DNA is required for a simple PCR and up to 20µg for gene sequencing analysis. DNA should be supplied in water or TE buffer at a concentration of ≥20µg/ml. Please see the Laboratory Test Directory for further information.

**Amniotic fluids for chromosome and DNA investigations (including QF-PCR)**

Families with DNA or cytogenetic abnormalities (i.e., hereditary genetic disorders or hereditary chromosome abnormalities) may require work-up prior to offering prenatal diagnosis. It is therefore important that the laboratory is alerted as early as possible when the specimen is expected for prenatal diagnosis requiring molecular DNA or cytogenetic investigations. Please telephone or email the laboratory as soon as possible to discuss, giving the patient details along with the nature of the genetic abnormality. Details should also be emailed to Clinical Genetics on GeneticsReferrals@ggc.scot.nhs.uk. There is no need to contact the laboratory or Clinical Genetics for routine QF-PCR testing.

**Two samples are required for amniotic fluid prenatal diagnosis**. For immediate processing, specimens must be received before 12.30pm or they may not be processed until the following day.

|  |  |
| --- | --- |
| **Specimen type** | **Specimen container** |
| 15-20mls amniotic fluid samples | Sterile 20ml universal  |
| 3ml maternal blood | EDTA (KE) specimen tube |

**Three samples are required for amniotic fluid prenatal diagnosis from pregnancies with an abnormal ultrasound scan**. For immediate processing, specimens must be received before 12.30pm or they may not be processed until the following day.

|  |  |
| --- | --- |
| **Specimen type** | **Specimen container** |
| 15-20mls amniotic fluid samples | Sterile 20ml universal  |
| 3ml maternal blood | EDTA (KE) specimen tube |
| 3ml paternal blood | EDTA (KE) specimen tube |

**Chorionic villus biopsy for chromosome and DNA investigations (including QF-PCR)**

Families with DNA or cytogenetic abnormalities (i.e., hereditary genetic disorders or hereditary chromosome abnormalities) may require work-up prior to offering prenatal diagnosis. It is therefore important that the laboratory is alerted as early as possible when the specimen is expected for prenatal diagnosis requiring molecular DNA or cytogenetic investigations. Please telephone or email the laboratory as soon as possible to discuss, giving the patient details along with the nature of the genetic abnormality. Details should also be emailed to Clinical Genetics on GeneticsReferrals@ggc.scot.nhs.uk. There is no need to contact the laboratory or Clinical Genetics for routine QF-PCR testing.

**Two samples are required for chorionic villus biopsy prenatal diagnosis.**

|  |  |
| --- | --- |
| **Specimen type** | **Specimen container** |
| 10 – 20 mg of villus for most referrals (see below) | Sterile 20ml universal containing transport medium (see below) |
| 3ml maternal blood | EDTA (KE) specimen tube |

**Three samples are required for chorionic villus biopsy prenatal diagnosis from pregnancies with an abnormal ultrasound scan.**

|  |  |
| --- | --- |
| **Specimen type** | **Specimen container** |
| 10 – 20 mg of villus for most referrals (see below) | Sterile 20ml universal containing transport medium (see below) |
| 3ml maternal blood | EDTA (KE) specimen tube |
| 3ml paternal blood | EDTA (KE) specimen tube |

The villus biopsy sample should be placed in a sterile universal containing sterile CVS transport medium (supplied by the laboratory, please contact us directly for further information). If additional testing is requested, for example for biochemical diagnosis, additional fetal material may be required and will require prior discussion with the laboratory before sampling.

For immediate processing, specimens must be received before 12.30pm or they may not be processed until the following day. Specimens received later in the day will miss the 12.30pm cut-off for QF-PCR testing but will be processed the following day.

**Fresh or frozen tissue specimens**

Products of conception or post-mortem samples should be sent to the laboratory in a dry well sealed container. Fresh tissue biopsies should be transported to the laboratory in sterile tissue culture medium (available on request), an internal tissue such as rib or pericardium is optimal for culture purposes. DNA can be extracted from fresh and frozen tissue specimens and the optimal tissue type for DNA extraction is spleen or lung. Frozen tissue specimens should be transported on dry ice. Fresh and frozen tissue specimens should be sent to the laboratory immediately after sampling.

**Other specimen types**

Venous blood sampling for molecular DNA investigations is the preferred specimen of choice, but occasionally this may not be appropriate. In these circumstances we may be able to extract DNA from other types of specimens such as buccal swabs or saliva samples, however other specimen types may yield insufficient DNA quality or quantity for some tests. ***Please contact the laboratory for further information prior to sampling for other specimen types.***

**RNA studies**

The extraction of RNA from some specimen types is available following discussion with the laboratory. ***Please telephone the laboratory prior to sampling for RNA studies.***

**DNA and RNA storage**

Extracted DNA/ RNA is retained for storage unless the request card indicates that permission for storage is denied. When storage is denied, the DNA/ RNA specimen is destroyed following testing. The laboratory can also store DNA and/ or RNA from patients on request (for example where no specific test is currently available).

# Specimen requirements for solid tumour investigations

**Fresh Tissue specimens**

Cytogenetics investigations (including FISH) from fresh tissue ideally require a 1cm piece of fresh tumour however, smaller pieces of tissue may be sufficient. Alternatively, a fine needle aspirate (FNA) or core biopsy with as much available material as possible should be sent for testing.

Fresh tissue samples should be placed in a sterile universal containing sterile tumour transport medium (supplied by the laboratory, please contact us directly for further information) and sent to the laboratory without delay.

Specimens taken on a Saturday or Sunday should be kept at room temperature and transported to the laboratory first thing on the Monday morning.

**FFPE tissue specimens**

Molecular investigations (DNA, RNA and FISH) from FFPE referrals should be performed using an identified tumour specific FFPE tissue block.

When molecular DNA/RNA investigations involving PCR are required, the identified tumour specific FFPE tissue block should be sent directly to the laboratory for sectioning to prevent cross contamination of the extracted DNA/ RNA specimen. The tumour cellularity (% tumour content) of the referred specimen **must** be detailed on the tumour request form. Targeted DNA extraction is available on request or is performed routinely when tumour load is low (e.g. <30% tumour versus normal tissue). The targeting process is performed in Laboratory Genetics in partnership with Pathology colleagues. Please contact the laboratory for further information.

**DNA and RNA storage**

Extracted DNA/ RNA is retained for storage unless the request card indicates that permission for storage is denied. When storage is denied, the DNA/ RNA specimen is destroyed following testing.

# Specimen requirements for haematological malignancy investigations

**Paediatric acute lymphoblastic leukaemia MRD specimens**

Minimal residual disease (MRD) testing is available at diagnosis and relapse using a blood (when WCC > 20x106) or a bone marrow specimen. For diagnostic testing either a blood or a bone marrow specimen is appropriate. For MRD monitoring at follow up time points only a bone marrow is acceptable. Specimens must be collected in ACD specimen tubes.

|  |  |
| --- | --- |
| **Specimen type** | **Specimen container** |
| 5 – 10ml blood (when WCC > 20x106) | ACD specimen tube  |
| 3 – 5ml bone marrow | ACD specimen tube |
| Trephine | Transport media |

There is some evidence that sample site and volume may affect MRD detection. For this reason careful attention to sampling technique is required. Please obtain marrow as follows: the aspirate needle should be passed fresh into two sites (this can be done by re-angling the needle; new skin punctures are not required). From each site the first 1ml of marrow should be aspirated using a 2ml syringe. The samples from each site can be combined into a single ACD specimen tube.

**Adult and paediatric haemato-oncology specimens**

Bone marrow is the preferred specimen for most haematological malignancy testing. Peripheral blood specimens may provide a result where there are circulating blast cells and/ or the white blood cell count is >20x109/l. Bone marrow trephine specimens are also acceptable, in cases of dry tap or where aspiration is difficult.

For cytogenetic karyotype and FISH investigations, specimens should be collected in bone marrow transport medium supplied by the laboratory. The specimens should be well mixed to prevent clotting and then transported to the laboratory **immediately** after collection. For the best results for patients, it is preferable for bone marrow samples to be received into the laboratory Monday to Thursday as the laboratory does not provide a Saturday service.

The transport medium is supplied in sterile 20ml universal tubes and should be stored at 4-8oC. Please do not use after the expiry date on the universal tube or if it appears cloudy/ contaminated.

When an urgent diagnosis is required (e.g. APML, CML), FISH can also be performed using an EDTA or LiHep blood specimen. **Please phone the lab on 0141 354 9320 before sending the specimen.**

For molecular DNA and RNA investigations, specimens should be collected in sterile tubes containing potassium EDTA (KE). The specimen should be well mixed to prevent clotting and then transported to the laboratory immediately after collection. It is essential that specimens which require RNA extraction arrive in the laboratory within 24 - 48 hours of sampling. Tests which involve RNA analysis may be compromised if the specimen arrives more than 48 hours after sampling.

|  |  |
| --- | --- |
| **Test required** | **Specimen type and notes** |
| **Karyotype & FISH** | * 1-3ml bone marrow (in supplied marrow medium)
* Or 5ml blood (LiHep)
* Or trephine (in supplied marrow medium)
 |
| **Karyotype, FISH & PCR** **(including RQ-PCR)\*** | * 1-3ml bone marrow (in supplied marrow medium)
* Or 5ml blood (LiHep)
* Or trephine (in supplied marrow medium)

Plus* 1-3ml bone marrow (EDTA)
* Or 5ml blood (EDTA) for routine PCR
* Or 20ml blood (EDTA) for RQ-PCR
* Or trephine (in supplied marrow medium)
 |
| **FISH only, CLL and Myeloma** | * CLL, 20ml blood (EDTA)
* Myeloma, 1-3ml bone marrow (EDTA)
 |
| **FISH urgent, CML and APML** | * 1-3ml blood (EDTA or LiHep or bone marrow)

***Follow up bone marrow required for karyotyping*** |
| **PCR alone** **(including RQ-PCR)** | * 1-3ml bone marrow (EDTA)
* Or 5ml blood (EDTA) for routine PCR
* Or 20ml blood (EDTA) for RQ-PCR
* Or trephine
 |

**\****Please note that all new diagnoses of acute myeloid leukaemia require both a cytogenetics sample for karyotype/FISH analysis and an EDTA sample for PCR analysis*

**Chimerism testing**

Blood samples are the preferred specimen for Chimerism studies, bone marrow can also be tested if required. Chimerism testing is available for both paediatric and adult samples.

|  |  |
| --- | --- |
| **Chimerism** | * Pre-transplant (donor and recipient), 5ml blood EDTA
 |
| * Post-transplant whole sample analysis, 5ml blood EDTA or 1-3ml bone marrow EDTA
 |
| * Post-transplant lineage specific analysis, 20ml blood EDTA or 1-3ml bone marrow EDTA
 |

**DNA and RNA storage**

Extracted DNA/ RNA is retained for storage unless the request card indicates that permission for storage is denied. When storage is denied, the DNA/ RNA specimen is destroyed following testing. DNA/ RNA for post-diagnostic acquired samples, i.e. monitoring specimens, is stored for 6 months.

# Testing for other disorders

Some of the genes or genetic abnormalities listed in the test directories may also be applicable to other disorders. Please contact the laboratory if you would like to discuss further testing.

Testing for other genetic disorders is also available from other Scottish regional centres through the SSNGM. For further information please contact these laboratories directly. Approved testing if unavailable in the West of Scotland Centre (Glasgow), will be forwarded on to an appropriate laboratory within SSNGM.

Contact details for the other Scottish Genetics Regional Laboratories:

<https://www.nss.nhs.scot/specialist-healthcare/specialist-services/genetic-and-molecular-pathology-laboratories/>