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Laboratory Information

The Service

The Biochemical Genetics Department comprises the West of Scotland Pregnancy Screening Service for Downs Syndrome and the Scottish Newborn Screening Laboratory and provides both a supra-regional and national screening service to Scotland. The laboratory is situated on level 2B of the Laboratory Medicine Building at the Queen Elizabeth University Hospital campus and along with Laboratory Genetics (Cytogenetic and Molecular Diagnostics) forms part of the West of Scotland Genetics Services.

The screening services provided are funded by National Services Division of NHS Scotland and are accredited as by Clinical Pathology Accreditation (CPA) UK Ltd (CPA Reference Number 2156).

Laboratory Mission Statement

To provide and further develop a high quality pregnancy and newborn screening services for those disorders where there is demand, using the most effective technological approaches whilst working within national and professional guidelines. To securely and confidentially store serum and blood spot samples and information from individuals using this service, and to ensure the best possible use of the available resources and expertise in the testing of these patients.
Contacting the Laboratory

Mailing Address for correspondence and samples

Biochemical Genetics Department  
West of Scotland Genetic Services  
Level 2B, Laboratory Medicine Building  
Queen Elizabeth University Hospital  
1345 Govan Road  
Glasgow  
G51 4TF

Other ways of contacting the laboratory

General Pregnancy Screening enquiries. Tel: 0141 354 9272 or 9273 (Internal 59272/3)  
General Newborn Screening enquiries  
Tel: 0141 354 9277 (Internal 59277)

Web:  http://www.nhsggc.org.uk/medicalgenetics (Internet)  
Staffnet pages - medical genetics. (NHSGGC intranet only)

Email:  GG-UHB.PrenatalScreeningLaboratory@nhs.net  
GG-UHB.NewbornScreeningLaboratory@nhs.net

These 2 email accounts are monitored daily and are suitable for the secure receipt of patient-identifiable information. Patient identifiable data may be transmitted to these either an @nhs.net or @”any”.scot.nhs.uk email address. Patient identifiable information should **NOT** be sent to the laboratory from any other email provider or to any other laboratory email address.

Department working hours

Monday to Friday 8.30a.m. – 5.00p.m.

The laboratory does not normally offer an out of hour’s service but it may be possible under exceptional circumstances, to arrange the analysis of urgent samples out-with these times, by prior arrangement.
Laboratory Staff

Head of Service
Gordon Lowther, Healthcare Scientist Consultant
Tel: 0141 354 9272
Email: gordon.lowther@ggc.scot.nhs.uk

Pregnancy Screening Coordinator
Louise Brown, Healthcare Scientist Principal
Tel: 0141 354 9274
Email: louise.brown@ggc.scot.nhs.uk

Newborn Screening Coordinator
Sarah Smith, Healthcare Scientist Principal
Tel: 0141 354 9275
Email: sarah.smith2@ggc.scot.nhs.uk

Laboratory Manager
Gary McCaw, Healthcare Scientist Principal
Tel: 0141 354 9275
Email: gary.mccaw@ggc.scot.nhs.uk

Quality Manager
Dr Gordon Graham, Healthcare Scientist Principal
Tel: 0141 354 9273
Email: gordon.graham@ggc.scot.nhs.uk

Other Senior Scientific Staff
Liz McBride, Healthcare Scientist Advanced
Tel: 0141 354 9272
Email: liz.mcbride@ggc.scot.nhs.uk

Moira Fitch, Healthcare Scientist Advanced
Tel: 0141 354 9278
Email: moira.fitch@ggc.scot.nhs.uk

Alison Estell, Healthcare Scientist Advanced
Tel: 0141 354 9278
Email: Alison.Estell@ggc.scot.nhs.uk

Other Useful Contact numbers
West of Scotland Clinical Genetics Department
Tel: 0141 354 9201

Laboratory Genetics Department (Cytogenetics And Molecular Diagnostics)
Tel: 0141 354 9330
Complaints

Should you have any comments, suggestions, cause for concern or complaints about the service you receive, please contact the Quality Manager or the appropriate Screening Coordinator using the contact details above.

Sending a sample to the Laboratory

Referral information

Newborn Bloodspot Screening

Bloodspot request forms, glassines and freepost envelopes are supplied to users by the department. These are available on request by telephoning 0141 354 9277 or by emailing a request to GG-UHB.NewbornScreeningLaboratory@nhs.net. All information on the referral form (bloodspot card) must be complete. Please note: - Scottish Government guidelines mandate that all Newborn Screening request forms received with a wrong or missing CHI number will be rejected and will require a repeat sample.

1st and 2nd Trimester Pregnancy Screening

Request forms are supplied to users by the department. These are available on request by telephoning 0141 354 9272 or by emailing a request to GG-UHB.PrenatalScreeningLaboratory@nhs.net. Referral forms must be legible and should contain the patient’s surname, forename, date of birth, CHI number, postcode and a name and address for the referring clinician for reporting purposes. The form should be fully completed with all maternal and pregnancy characteristics as requested.

Each request accepted by the laboratory for testing is considered an agreement between the user and laboratory.

Consent

All screening tests require appropriate consent. The laboratory assumes that by sending a sample consent has been obtained by the referring clinician (i.e. implied consent). For further information and guidance, please refer to the appropriate NS Screening guidelines at:-

http://www.nsd.scot.nhs.uk/services/screening/index.html

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.
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**.

**High risk specimens**

If a sample is known to present a high-risk to laboratory staff e.g. where the patient is affected with HIV, Hepatitis B or Hepatitis C, then the referral card and the specimen tube should be clearly labelled as ‘danger of infection’ or ‘high risk’. The laboratory **cannot** process specimens from patients who have or who are suspected of having Group 4 pathogens.

**Packaging and transportation**

The sending of pathological specimens by taxi, through the post or by courier is subject to specific regulations. 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. Further information regarding the packaging and transport of specimens can be found at, [http://www.unece.org/fileadmin/DAM/trans/danger/publi/adr/adr2013/English/Volume1.pdf](http://www.unece.org/fileadmin/DAM/trans/danger/publi/adr/adr2013/English/Volume1.pdf) and [http://www.royalmail.com/sites/default/files/Guidance-Document-Infectious-Substances-171012.pdf](http://www.royalmail.com/sites/default/files/Guidance-Document-Infectious-Substances-171012.pdf).

Whole Blood 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.

Further details of local sampling and transportation guidelines can be found under the appropriate Departmental sections of this handbook.

**Biochemical Genetics Service Departments**

The Biochemical Genetics Department is sub-divided into two sections:

*Prenatal Screening, Newborn Screening*

Enquiries regarding testing, results and advice should be directed to the Screening Coordinator of the appropriate section.

**Prenatal Screening for Neural Tube Defects and Down’s Syndrome.**

This service is provided for 60% of the Scottish pregnant population in seven Health Boards in the West of Scotland: Ayrshire and Arran, Dumfries and Galloway, Forth valley, Greater Glasgow and Clyde, Lanarkshire, Highland and the Western Isles.
Samples are also accepted from further afield. In the west of Scotland, over 20,000 women choose to have the prenatal screening test for Down's syndrome each year. Screening for Down's syndrome started in 1991 and to date, this is one of the largest prenatal screening programmes in Europe.

**Test principle:**

**First Trimester Screening (Combined Ultrasound and Biochemical Screening):** This uses a combination of ultrasound nuchal translucency measurements at 11+2 to 14+1 weeks of gestation and the serum biochemical markers, free beta hCG and Pregnancy Associated Plasma Protein A, (PAPP-A). For further information, request forms and patient information leaflets, contact the Prenatal Screening Office (0141 354 9272 / 9273). Please use the green request form for 1st trimester screening.

Women who have received a Down’s risk in the first trimester should not have a second trimester sample taken. Neural Tube Defects should be detected by ultrasound anomaly scanning.

**Second Trimester Screening:** The concentrations of four maternal serum markers, alpha-fetoprotein (AFP), human chorionic gonadotrophin (hCG), Inhibin A (InhA) and unconjugated estriol (UE3) are measured in maternal blood at 14+2 to 20+0 weeks of gestation. Please use the white request form for second trimester screening.

A probability (risk) that each analyte level is associated with a Down's syndrome pregnancy is derived and combined with the prior probability of an affected pregnancy based on maternal age. Women with a combined risk of $\geq 1$ in 150 are considered to be at increased risk and are offered diagnostic testing.

**Sample requirements:** For Down’s Syndrome screening a clotted venous blood sample (5-10mls) is required. Note: Blood should be collected in plain (serum) tubes as some anticoagulants e.g. EDTA interfere with the biochemical test.

All sample tubes must be labelled and dated, and sealed in the bag attached to the appropriate completed request form bearing the CHI number. All clinical information should be filled in as this is essential for accurate interpretation of test results. Patient information leaflets are available from the local Health Board Health Promotion Department. Leaflets in languages other than English can be obtained via the internet at:


**Sending samples to the Laboratory:** Samples should be sent to the laboratory with minimum delay after being taken. Samples received more than 3 days after the sample date are no longer suitable for analysis for first trimester testing (5 days for second trimester testing) and a repeat sample will be requested. Sample tubes must be sealed into the sample bag attached to the request form. Samples forwarded by post must be packaged to conform to current postal regulations and should be sent first class/guaranteed next day delivery.

**Results:** Over 95% of results are available within 3 working days of sample receipt. Results which require diagnostic follow-up testing for Down's syndrome are emailed to a secure address in each of the referring centres, as soon as the result is available. Hard copies of reports are generated for all samples and sent to the referring centre by mail.
For further information on prenatal screening see the Biochemical Genetics pages at: http://www.nhsggc.org.uk/medicalgenetics

Scottish Newborn Screening Laboratory

The remit of the Scottish Newborn Screening Laboratory (SNSL) is to screen all babies born in Scotland (currently around 60,000 per annum) for phenylketonuria (PKU), congenital hypothyroidism (CHT), cystic fibrosis (CF) Sickle Cell Disorders (SCD) and Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD).

The aim of the service is to diagnose these disorders as early as possible to allow affected infants to be placed on the appropriate treatment.

Clinical advice.

Phenylketonuria

About 1 in 8,000 babies born in Scotland has phenylketonuria (PKU). Babies with this inherited condition are unable to process a substance in their food called phenylalanine. If untreated, they will develop serious, irreversible, mental disability. Screening means that babies with the condition can be treated early through a special diet, which will prevent severe disability and allow them to lead a normal life. If babies are not screened, but are later found to have PKU, it may be too late for the special diet to make a real difference.

Congenital hypothyroidism

About 1 in 3,500 babies born in Scotland has congenital hypothyroidism (CHT). Babies with CHT do not have enough of the hormone thyroxine. Without this hormone, they do not grow properly and can develop serious, permanent, physical and mental disability. Screening means that babies with CHT can be treated early with thyroxine tablets, which will prevent serious disability and allow them to develop normally. If babies are not screened and are later found to have CHT, it may be too late to prevent them becoming seriously disabled.

Sickle cell disease

About 1 in 2,500 babies born in the UK has a sickle cell disease (SCD). These are inherited disorders that affect the red blood cells. If a baby has a sickle cell disease, their red blood cells can change to a sickle shape and become stuck in the small blood vessels. This can cause pain and damage to the baby’s body, serious infection, or even death. Screening means that babies with SCD can receive early treatment, including immunisations and antibiotics, which, along with parent education, will help prevent serious illness and allow the child to live a healthier life.

Cystic fibrosis
About 1 in 2,500 babies born in Scotland has cystic fibrosis (CF). This inherited condition can affect the digestion and lungs. Babies with CF may not gain weight well, and have frequent chest infections. Screening means that babies with CF can be treated early with a high energy diet, medicines and physiotherapy. Although a child with CF may still become very ill, evidence suggests that early treatment helps them live longer healthier lives.

**MCADD (Medium-Chain Acyl-coA Dehydrogenase Deficiency)**

About 1 in 10,000 babies born in Scotland has MCADD. Babies with this inherited condition have problems breaking down fats to make energy for the body. This can lead to serious illness, or even death. Screening means that most babies who have MCADD can be recognised early, allowing special attention to be given to their diet, including making sure they eat regularly. This care can prevent serious illness and allow babies with MCADD to develop normally. Screening babies for MCADD is important because it enables those with the condition to be identified before they become suddenly and seriously ill.

If a family history of any of these disorders is known, a clinician may arrange testing at birth. It is important to remember that the routine screen at day 5-8 will still be needed.

**Will screening for these conditions show up anything else?**

Screening for cystic fibrosis (CF) includes testing some babies for the most common gene alterations that cause CF. This means screening may identify some babies who are likely to be genetic carriers of cystic fibrosis. These babies may need further testing to find out if they are a healthy carrier, or have CF. Screening identifies babies who are genetic carriers of sickle cell or other unusual red blood cell disorders. Carriers of sickle cell disorders are healthy and will not be affected by the condition. Rarely, other serious conditions such as beta thalassaemia major can be identified. In this condition, the baby does not make enough red blood cells, and needs treatment for severe anaemia. The newborn screening lab will always refer a baby to a specialist clinician if a serious condition is detected.

More specific clinical advice on the screening programmes and on these conditions can be obtained by contacting the laboratory on 0141 354 9275 and we can direct your query to the most appropriate member of staff.

**Sample requirements:**

The routine dried blood spot specimen should be collected when the baby is at least 96 hours old and ideally on day 5 of life.

A good quality blood sample is one that:

- is taken at the right time
- is appropriately labelled to allow identification of the baby, analysis and reporting of results
- contains sufficient blood to perform all tests i.e. blood has soaked through to the back of the card
- has not been contaminated or taken on an expired blood spot card
• arrives at the laboratory in a timely manner

For further information on the guidelines regarding blood spot collection please follow the link below:

http://www.nsd.scot.nhs.uk/services/screening/newbornscreening/

A summary of the instructions for collecting the blood spot specimen are printed on the reverse of the blood spot card. Briefly, the heel is swabbed, warmed and punctured with a suitable bloodletting device and the blood droplets applied to the filter paper portion of the blood collection card. All details requested on the card should be filled in at the time of collection. Blood spot cards can be obtained on request from the laboratory.

Transporting the Sample

The blood spot card should be air dried flat in a horizontal position and then placed in a glassine envelope provided with the card and sent directly to the SNSL in the pre-addressed, free post envelope as soon as possible after collection. Only one blood spot card should be placed in each glassine envelope. No more than six blood spot cards should be placed in a single envelope as this will exceed the contracted postal weight and delay the delivery of specimens.

In the event of a postal strike, staff should refer to their local NHS Board protocols and contingency plans to ensure the timely delivery of blood spot cards to the SNSL.

Factors that can affect the test results

• Specimen taken too early (before 96 hours of age)
• Specimen taken too soon after a transfusion (less than 72 hours post transfusion)
• Specimen taken on an expired blood spot card
• Specimen delayed in transit i.e. it has taken more than 14 days from specimen collection to receipt in the laboratory
• There is insufficient sample to perform all tests.
• There is incomplete information on the card e.g. date of specimen is not included.
• Specimen has not been dried properly prior to transit.

Patient information leaflets are available from the local Health Board Health Promotion Department. An electronic version, including leaflets in languages other than English, can be obtained via the internet:


Test Principle:

Dried blood spots from newborn babies are tested by mass spectrometry for high levels of Phenylalanine (as an indicator of PKU) and Octanoylcarnitine (as an indicator of MCADD). Delfia immunoassay is used to measure Thyroid Stimulating Hormone (TSH), as an indicator of Hypothyroidism and immunoreactive trypsinogen (IRT) as an indicator of CF. DNA is extracted from blood spots from newborn babies with elevated concentrations of IRT and analysed for pathogenic mutations. High performance liquid
chromatography (HPLC) is used as the primary screen for SCD with Isoelectric focusing (IEF) used as a confirmatory method.

Blood spot specimens that require molecular analysis are forwarded to:

Laboratory Genetics,
West of Scotland Genetic Services,
Laboratory Medicine,
Queen Elizabeth University Hospital
Glasgow G51 4TF

**Results:** The SNSL reports blood spot screening results to the relevant NHS Board Child Health/Screening Department where a record is maintained of all babies resident in the area. If a repeat sample is required, the SNSL notifies the relevant Child Health/Screening Department to arrange for a repeat sample to be obtained by the midwife or health visitor.

**Turnaround times:** The SNSL will report all suspected results on the day of confirmation within the laboratory to the nominated clinical specialist. The SNSL aims to have 95% of all written results reported within two working days of receipt of the specimen arriving within the laboratory.

More information on Newborn Screening and the West of Scotland Genetic Services can be found at: [http://www.nhsggc.org.uk/medicalgenetics](http://www.nhsggc.org.uk/medicalgenetics)

**Improvements to the service**

We aim to provide a high quality service and are constantly striving to improve. In order to do this we value any constructive comments from our users. If you have any comments you would like to make please feel free to contact our Quality Manager.

Dr Gordon Graham, Principal Clinical Scientist
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Email: gordon.graham@ggc.scot.nhs.uk