

South West Laboratory Service for Newborn Screening and Metabolic Biochemistry, Southmead Hospital, Bristol

Handbook for users



Please check this is the current version of the user handbook at:

www.nbt.nhs.uk/severn-pathology/pathology-services/clinical-biochemistry/metabolic-biochemistry

For more information on our pathology services please visit:

www.nbt.nhs.uk/severn-pathology

Exceptional healthcare, personally delivered

Contents

1. Introduction to Newborn Screening and Metabolic Biochemistry.....	3
Newborn Screening	3
Metabolic Biochemistry	3
Clinical or Laboratory Advisory Service.....	4
2. Service Standards/Quality Assurance.....	4
Service Commitment.....	4
3. Useful Contacts	5
4. Service Location & Availability	6
Service Hours	6
Address of the Department	6
5. Sending a Specimen for Newborn Screening	6
6. Metabolic Biochemistry - Specimen Collection, completion of the request form and management of urgent and additional requests	7
Sending a Specimen for Metabolic Biochemistry.....	7
Instructions for patient collected samples.....	7
Instructions for the completion of the request form	7
Specimen Labelling.....	8
Urgent Samples	9
7. Analyses offered by the laboratory	9
8. Guidance on Galactosaemia Investigations.....	14
9. Work Referred Away	17
10. Information Governance.....	17
Privacy Policy and Data Protection	17
How we handle your information	17

1. Introduction to Newborn Screening and Metabolic Biochemistry

The South West Laboratory service for Newborn Screening and Metabolic Biochemistry is provided by the department of Clinical Biochemistry located within the Blood Sciences Laboratories at Southmead Hospital.

Newborn Screening

The Newborn Screening Laboratory provides the Newborn Bloodspot Screening service for a large part of the South West Region. The Laboratory is a stakeholder Laboratory in UK Newborn Screening Laboratory Network (UKNSLN). Testing is undertaken on filter paper bloodspots which are collected at 5 days of age. The laboratory currently screens approximately 36,000 babies each year for nine conditions:

- Sickle cell disease (SCD)
- Cystic Fibrosis (CF)
- Congenital Hypothyroidism (CHT)
- Inherited Metabolic Diseases:
 - Phenylketonuria (PKU)
 - Medium-chain acyl-CoA dehydrogenase deficiency (MCADD)
 - Maple syrup urine disease (MSUD)
 - Isovaleric acidaemia (IVA)
 - Glutaric aciduria type 1 (GA1)
 - Homocystinuria (pyridoxine unresponsive) (HCU)

Early detection through screening and prompt treatment significantly reduces the morbidity and mortality associated with these conditions. The newborn screening bloodspot programme is provided in close collaboration with health care professionals throughout the region.

Please refer to the separate Public Health England newborn screening handbook for further details:

<https://www.gov.uk/government/publications/health-professional-handbook-newborn-blood-spot-screening>

Metabolic Biochemistry

The Metabolic Biochemistry Laboratory provides a regional diagnostic and monitoring service for the investigation of inherited metabolic diseases and is a Stakeholder Laboratory of the UK National Metabolic Biochemistry Network (MetBioNet). The laboratory professionals work closely with the SW Regional Paediatric Metabolic Team based at the Bristol Royal Hospital for Children and Adult IMD teams based at North Bristol NHS Trust and United Hospitals Bristol and Weston NHS Foundation Trust.

Routine analyses performed include urine organic acids (including quantitative methylmalonic acid, if required), bloodspot/plasma acylcarnitines (including free carnitine), plasma/urine/CSF quantitative amino acids, qualitative galactosaemia screening test, plasma free fatty acids and 3-hydroxybutyrate.

In addition, the laboratory provides specialist testing for the investigation of peroxisomal disorders (plasma very long chain fatty acid analysis, including pristanic and phytanic acids), and Smith-Lemli-Opitz syndrome (plasma 7-dehydrocholesterol and 8-dehydrocholesterol).

The laboratory is a centre for galactosaemia investigations and can offer a range of assays for the investigations of disorders of galactose metabolism.

- Quantitative galactose-1-phosphate uridylyltransferase activity
- Red Cell Galactose-1-phosphate
- Red Cell galactokinase activity
- Urine galactitol

The department also measures bloodspot 17-OHP to monitor patients with Congenital Adrenal Hyperplasia, and bloodspot TSH for monitoring patients with Downs syndrome or needle-phobia to screen for primary hypothyroidism or to detect under-replacement with thyroxine.

Clinical or Laboratory Advisory Service

We provide a comprehensive advisory service and welcome enquiries to discuss appropriate patient investigation and result interpretation. Clinical and laboratory advice is available during working hours.

For routine enquiries, urgent analyses or additional testing on samples already received, please telephone the laboratory to discuss. See the 'Contact the laboratory' for further information.

2. Service Standards/Quality Assurance

The department is fully accredited by the United Kingdom Accreditation Service to ISO15189. Our accreditation covers those activities described in our Schedule of Accreditation, which can be found at www.nbt.nhs.uk/severn-pathology/quality/pathology-accreditation-status

The quality of our service is maintained by recognised effective internal quality control measures and by participation in the External Quality Assessment (EQA) schemes provided by UKNEQAS, ERNDIM and CDC. Where schemes are not available, we participate in interlaboratory comparison schemes with other laboratories where possible. The staff working within the department are fully qualified, HCPC registered (where required), specialised and experienced. A high-quality service is maintained by frequently looking at feedback from user meetings, audits, and user

satisfaction surveys. Our Quality Policy and Manual contain further information and are available upon request.

Newborn Screening and Metabolic Biochemistry within Clinical Biochemistry at North Bristol NHS Trust is approved for Biomedical and Clinical Scientist training by the Health and Care Professions Council (HCPC) and by the Royal College of Pathologists and Royal College of Physicians for Chemical Pathology and Metabolic Medicine training. The Department also provides many educational activities for its own staff and for a range of healthcare professionals including medical students, junior doctors and local GPs and plays an active role in Clinical Audit, Research and Development in a wide variety of areas.

Tests not performed within the Trust may be sent to Supra Regional Services or other approved laboratories.

Service Commitment

The purpose of this handbook is to provide information on the South West Newborn Screening and Metabolic Biochemistry laboratory service including test repertoire, specimen requirements and details on accessing our service. It also provides some guidance on investigating patients for suspected Inherited Metabolic Disorders.

3. Useful Contacts

Hospital switchboard: 0117 950 5050 Duty Biochemist: 0117 4148437

Further information can be found on our website: www.nbt.nhs.uk/severn-pathology

Dr Helena Kemp (Consultant Chemical Pathologist/Director of Newborn Screening) helena.kemp@nbt.nhs.uk 0117 414 8425

Nicola Crabbe (Lead Biomedical Scientist for Specialist Biochemistry) nicola.crabbe@nbt.nhs.uk 0117 414 8450

Metabolic Biochemistry section

Result enquiries (email) NBS&MetabolicBiochemistry@nbt.nhs.uk

Result enquiries (telephone) 0117 414 8346

Maryam Khan (Principal Clinical Scientist) maryam.khan@nbt.nhs.uk 0117 414 8418

Bryony Wright (Senior Biomedical Scientist) bryony.wright@nbt.nhs.uk 0117 414 8346

Newborn Screening section

Result enquiries (email) NewbornScreening@nbt.nhs.uk

Result enquiries (telephone and answering machine 09:00-17:00) 0117 414 8412

Emma Smith-Thomas (Senior Clinical Scientist) emma.smith-thomas@nbt.nhs.uk 0117 414 8427

Joanna Harding (Senior Clinical Scientist) joanna.harding@nbt.nhs.uk 0117 414 8439

Clare Le Masurier (Senior Biomedical Scientist) clare.lemasurier@nbt.nhs.uk 0117 414 8430

4. Service Location & Availability

Service Hours

Normal Working Hours

The laboratories are open for the receipt and processing of routine samples and for result reporting and advice from 09:00-17:00 Monday to Friday (except Bank Holidays).

Out of Hours Service

At NBT within clinical Biochemistry we provide an out of hours service to analyse plasma ammonia and CSF and plasma lactate if required.

If analytical or advisory services are required out of hours, please contact the on-call Consultant Biochemist in the first instance.

Address of the Department

South West Newborn Screening and Metabolic Biochemistry Laboratory

Pathology Sciences Building

Southmead Hospital

North Bristol NHS Trust

Bristol

BS10 5NB

5. Sending a Specimen for Newborn Screening

Newborn screening has a dedicated PO Box to enable the post to get to the lab as quickly as possible. Transport links between Screening areas and the lab vary between maternity units, if you are in doubt about this procedure, please contact your local screening co-ordinator.

Newborn screening PO address for post and Couriers

Newborn Screening Laboratory

Pathology Sciences Laboratory

PO Box 407

BRISTOL

BS9 0EA

6. Metabolic Biochemistry - Specimen Collection, completion of the request form and management of urgent and additional requests

Consent

Unless written consent is required for a particular test or investigation (this should be documented in the test details), the laboratory assumes that informed consent for testing to be carried out has been given at the time the request form has been completed. It is the responsibility of the requesting clinician to obtain consent for specimen collection and the tests requested. It is implicit in the receipt of the request form that consent has been obtained. This applies to all Trusts sending samples.

Sending a Specimen for Metabolic Biochemistry

Information on sending samples collected within North Bristol NHS Trust can be obtained by following this link: www.nbt.nhs.uk/severn-pathology/requesting/transport.

Specimens collected at sites outside North Bristol NHS Trust should be sent via the Blood Sciences/Clinical Chemistry department in the requesting hospital. Many of these transport links are already in place. In some cases, specimens for certain tests may require immediate transport by courier or taxi. Specific needs are listed in the specimen requirement section of the table in this handbook. If using a courier or taxi, please request that the specimens are delivered to the South West Newborn Screening and Metabolic Biochemistry Laboratory. Please also try to avoid sending samples in an envelope instead use a sample box. All specimens must be handled with care and treated by all personnel as a potential infection risk.

However, additional precautions are required for samples that are deemed to be high risk.

There is a large volume of post sent to the Pathology Sciences Building and an envelope can sometimes be confused with paperwork/reports instead of blood samples.

- For details on individual specimen requirements see table below.
- For details on completing request forms and minimum criteria for a request see below.
- All received separate specimens are assumed to be from the correct sample type and requirements listed in the table below and will be interpreted accordingly.

Instructions for patient collected samples

For specimens collected by patients and/or their parents/carers (e.g., urine or bloodspot/capillary blood samples), instruction leaflets, and training are available and provided on request. For patients who need bloodspot cards for PKU monitoring or 17-OHP monitoring please contact the Newborn Screening office, see contacts list.

Instructions for the completion of the request form

It is the policy of North Bristol NHS Trust that all samples taken for laboratory investigations and accompanying request forms will be labelled to a minimum standard which minimises the risk of harm to patients. The Services Specimen Labelling Policy is accessible from our website www.nbt.nhs.uk/severn-pathology/requesting Key points are:

- The patient's primary identifier is the NHS number, and this must be used wherever possible.

- In normal circumstances, unlabelled or inadequately labelled samples will not be processed. There is a recognition that there are certain circumstances where samples are unrepeatable. The circumstances and actions in this case are documented within the policy.
- Each request received by the laboratory is considered an agreement between the requestor and laboratory for provision of Pathology Services.

Local Requests can be made use Sunquest ICE or externally by Labgnostic. Where this is unavailable, dedicated NBT Pathology Request forms are available which should be completed in full, as below. If the request is from outside of the Hospital trust, please ensure full compliance with the following.

ESSENTIAL CRITERIA ON REQUEST FORM
NHS number or other unique identity number
Patient Surname
Patient First name
Date of birth
Date and time of specimen collection & type of specimen
Investigations required
Name and location of requesting practitioner/contact number

DESIRABLE CRITERIA ON REQUEST FORM
Clinical information and medication where relevant
Purchase order number (external requests only)

Clinical information will help with interpretation of results therefore please include relevant details.

Specimen Labelling

ESSENTIAL CRITERIA ON SAMPLE
NHS number or other unique identity number
Patient Surname
Date of birth

DESIRABLE CRITERIA ON SAMPLE
Date and time of specimen collection

Neonatal Samples

It is recognised that labelling samples from neonates will require adaptation due to the potential lack of information available for a newborn and the small label size on neonatal bottles:

- Specimens must be labelled with infant's surname, date of birth and hospital number – this will be generally in the form of a printed label attached to the bottle.
- Request forms must be labelled as stated previously except that there may be no first name and the Hospital number may be used as the unique identity number.
- For multiple births, where there is no first name, specimens and request forms must clearly indicate the individual baby's status in addition to the surname, date of birth and Hospital number e.g., Twin 1, Twin 2.

Obtaining blood samples from neonates is very challenging and that as extreme pre-term infants have a very small circulating volume repeated sampling may be harmful. If there are concerns about the labelling of a sample the laboratory will contact the specimen collector to discuss the request.

Urgent Samples

Urgent requests can often be dealt with more quickly if there is prior discussion with the team. If your request is urgent, please contact the clinical team or the Duty Biochemist on the Contacts page.

Specimen Storage

Plasma and urine samples are stored frozen for 3 months prior to disposal. Newborn Screening bloodspot samples are stored indefinitely currently pending guidance from the Newborn Screening Bloodspot Screening programme on the retention and storage of bloodspots. On occasions it is necessary to want additional tests performed on the Newborn Screening card for example CMV testing. For information on this procedure please see

www.nbt.nhs.uk/severn-pathology/pathology-services/clinical-biochemistry/newborn-screening/blood-spot-retrieval-additional-tests

Add on tests

To request additional tests on samples already received by the laboratory, please telephone to discuss whether sufficient specimen is remaining and is suitable for analysis. For requests added after receipt of the specimen, turnaround times are from the date of test request. For further information please see www.nbt.nhs.uk/severn-pathology/requesting/test-information

Specimen rejection

Samples or request forms that do not meet the minimum labelling criteria may be rejected. Samples that are insufficient or have been collected incorrectly, for example in an incorrect preservative or received too late after sampling time for analysis will also be rejected. We do not routinely analyse these sample in Metabolic Biochemistry. If appropriate these unsatisfactory samples will be stored in the lab for 1 month.

7. Analyses offered by the laboratory

This section of the handbook explains which examinations are offered by the laboratory, including (as appropriate) information concerning samples required, sample volumes, special precautions, biological reference intervals and clinical decision values.

Testing Guidelines

The National Metabolic Biochemistry Network provides information about testing and guidelines for metabolic disorders:

<https://metbio.net/best-practice-guidelines/>

Test Repertoire

If the test you require is not included in our repertoire, please contact us, or visit the National Metabolic Biochemistry Website:

<https://metbio.net/resources/assay-directory/>

Test Cost

Available upon request

Test Name (Methodology)	Sample Volume, Type and Requirements	Special Instructions	Clinical Utility	Storage and Transport	Turnaround Times
7-Dehydrocholesterol (GCMS)	0.5ml Plasma (lithium heparin)	Should be protected from light	Diagnosis and monitoring of Smith-Lemli-Opitz (SLO) syndrome	Store and send frozen, if possible. Otherwise send room temperature by 1 st class post, unless courier arrangements are in place.	28 days
Amino Acids (Fully Quantitative) (Biochrom ion exchange chromatography)	0.5ml Plasma (lithium heparin)	Blood is the preferred sample type for investigation of the majority of disorders of amino acid metabolism; preprandial or fasting plasma sample preferred	Diagnosis and monitoring of disorders of amino acid metabolism; monitoring of patients with other known IMDs	Store and send frozen, if possible. Otherwise send room temperature by 1 st class post, unless courier arrangements are in place.	21 days
	0.5ml CSF (includes glycine, threonine, serine, alanine)	A paired plasma sample is required with CSF for interpretation.	Diagnosis of nonketotic hyperglycinaemia (NKH), and disorders of serine biosynthesis.		
	0.5ml Urine (no preservative)	If investigating renal stones for cystinuria, request random urine cystine	Diagnosis of disorders of amino acid transport (including Hartnups, lysinuric protein intolerance), Fanconi syndrome and hypophosphatasia (phosphoethanolamine)		
Acylcarnitine Bloodspot profile (includes free carnitine) (Tandem MSMS)	Bloodspots - 2 adequately sized spots and fully permeated the card. Please note we do not accept EDTA samples.	Please ensure Acylcarnitine is written in the comments section if using a Newborn Screening bloodspot card. Not to be sent with Newborn Screening post	Diagnosis and monitoring of disorders of fatty acid oxidation and ketone metabolism, and organic acidaemias	Ideally send sample in a glassine sleeve. Room temperature by 1 st class post, unless courier arrangements are in place.	21 days

Test Name (Methodology)	Sample Volume, Type and Requirements	Special Instructions	Clinical Utility	Storage and Transport	Turnaround Times
Plasma Carnitine and Acylcarnitine profile (includes free carnitine) (Tandem MSMS)	0.5ml Plasma (lithium heparin)	Please note we do not accept EDTA samples.	Diagnosis and monitoring of disorders of fatty acid oxidation and ketone metabolism, and organic acidaemias; preferred sample type for monitoring carnitine status/ketogenic diet	Store and send frozen, if possible. Otherwise send room temperature by 1 st class post, unless courier arrangements are in place.	21 days
Acylcarnitine Post-mortem Blood and Bile profile (Tandem MSMS)	Bloodspot/Bile - 2 adequately sized spots and fully permeated the card. Please note we do not accept EDTA samples.	Please ensure we have pathologist details on form and specify if post-mortem or peri-mortem.	Investigation of SUDI	Ideally send sample in a glassine sleeve. Room temperature by 1 st class post, unless courier arrangements are in place.	42 days
24 Hour Cystine excretion (Biochrom ion exchange chromatography)	0.5ml Urine (in HCL)	Time and date of collection and sample volume required.	Diagnosis of cystinuria	Store and send frozen, if possible. Otherwise send room temperature by 1 st class post, unless courier arrangements are in place.	21 days
Cystine (Day or Night) (Biochrom ion exchange chromatography)	0.5ml Urine (in HCL)	Time and date of collection and sample volume required.	Monitoring of cystinuria	Store and send frozen, if possible. Otherwise send room temperature by 1 st class post, unless courier arrangements are in place.	21 days
Combined urine day and night urine Cystine samples (Biochrom ion exchange chromatography)	0.5ml Urine (in HCL)	Time and date of collection and sample volume required.	Monitoring of cystinuria	Store and send frozen, if possible. Otherwise send room temperature by 1 st class post, unless courier arrangements are in place.	21 days
Random urine Cystine (Quantitation by Biochrom ion exchange chromatography)	0.5ml Urine (no preservative)	N/A	Screening for cystinuria	Store and send frozen, if possible. Otherwise send room temperature by 1 st class post, unless courier arrangements are in place.	21 days

Test Name (Methodology)	Sample Volume, Type and Requirements	Special Instructions	Clinical Utility	Storage and Transport	Turnaround Times
Free Fatty Acids & 3-hydroxybutyrate (Randox)	0.5ml Plasma (fluoride oxalate preferred) We do not accept heparinised samples	Free fatty acids unstable so should be sent to the laboratory frozen if possible. Please provide concurrent plasma glucose result for interpretation.	Investigation of hypoglycaemia, monitoring of ketogenic diet or prolonged fast	Store and send frozen, if possible. Otherwise send room temperature by 1 st class post, unless courier arrangements are in place.	7 days
Galactitol (GCMS)	0.5ml Urine (no preservative)	This test can be requested if classical galactosaemia is suspected in a transfused baby.	Screening for or monitoring disorders of galactose metabolism	Store and send frozen, if possible. Otherwise send room temperature by 1 st class post, unless courier arrangements are in place.	28 days
Galactosaemia screen (Beutler)	1ml Lithium heparin whole blood (received within 7 days of collection) or bloodspot.	This test is not valid if the patient has been transfused. Please do not send on a Friday.	Screening for classical galactosaemia	Refrigerated prior to sending samples. Whole blood shipped at room temperature/chilled. Should arrive within 7 days.	4 days
Galactose-1-phosphate (Tandem MS)	1ml Lithium heparin whole blood.	Please do not send on a Friday.	Confirmation and monitoring of classical galactosaemia	Refrigerated prior to sending samples. Whole blood shipped at ambient temperature/chilled arrive within 56 hours from sampling.	28 days
Galactokinase Quantitation (Tandem MS)	1ml Lithium heparin whole blood	Contact the laboratory prior to taking the sample.	Diagnosis of galactokinase deficiency	Whole blood shipped with ice pack/chilled to be assayed within 5 days of sampling	28 days
Galactose-1-phosphate uridyl transferase Quantitation (Tandem MS)	1ml Lithium heparin whole blood	Test not suitable if the patient has received any red cells within 4 months. Please do not send on a Friday.	Confirmation of classical galactosaemia, determination of carrier status in parents	Whole blood shipped at ambient temperature/chilled arrive within 48 hours from sampling.	28 days
Methylmalonic Acid Quantitation (GCMS)	1ml Urine (no preservative)	N/A	Investigation of B12 deficiency, monitoring of known IMD patients	Store and send frozen, if possible. Otherwise send room temperature by 1 st class post, unless courier	28 days

Test Name (Methodology)	Sample Volume, Type and Requirements	Special Instructions	Clinical Utility	Storage and Transport	Turnaround Times
				arrangements are in place.	
Organic Acids (GCMS)	5ml (minimum 2ml) Urine (no preservative)	Please alert laboratory if urgent analysis is required.	Investigation of suspected IMD, and diagnosis of primary organic acidurias and fatty acid oxidation disorders; includes detection of pyroglutamic acid and orotic acid	Store and send frozen, if possible. Otherwise send room temperature by 1 st class post, unless courier arrangements are in place.	14 days
Phytanic acid (GCMS)	0.5ml Plasma (lithium heparin) preferred Serum accepted	N/A	Monitoring of known patients with Refsum's disease	Store and send frozen, if possible. Otherwise send room temperature by 1 st class post, unless courier arrangements are in place.	21 days
Very Long Chain Fatty Acids (includes pristanic and phytanic acids) (GCMS)	0.5ml Plasma (lithium heparin) preferred Serum accepted	Adults – fasting sample strongly preferred. Infants – preprandial/pre feed sample recommended	Diagnosis of disorders of peroxisomal fatty acid oxidation and peroxisome biogenesis	Store and send frozen, if possible. Otherwise send room temperature by 1 st class post, unless courier arrangements are in place.	21 days

Test Name (Methodology)	Sample Volume, Type and Requirements	Special Instructions	Clinical Utility	Storage and Transport	Turnaround Times
17-Hydroxyprogesterone (17-OHP) (solid phase, time resolved fluoroimmunoassay)	Bloodspot - 1 adequately sized spot, fully permeated the card per time point.	Please ensure our specific request form and filter paper is used for this test. Contact the laboratory to order sample collection kit.	Monitoring of congenital adrenal hyperplasia (CAH)	Should arrive within 10 days of sample collection. Send room temperature by 1 st class post, unless courier arrangements are in place.	21 days

Test Name (Methodology)	Sample Volume, Type and Requirements	Special Instructions	Clinical Utility	Storage and Transport	Turnaround Times
Thyroid Stimulating Hormone on Bloodspot (TSH) (solid phase, time resolved fluoroimmunoassay)	Bloodspot - 2 adequately sized spots and fully permeated the card.	Mark the card with 'Bloodspot TSH' clearly, include a request form with details of where the report should be sent.	Monitoring children and adults with Down's syndrome or needle phobia, to screen for or monitoring of primary hypothyroidism	Should arrive within 10 days of sample collection. Send room temperature by 1 st class post, unless courier arrangements are in place.	7 days

Other specialist bloodspot tests offered:

8. Guidance on Galactosaemia Investigations

		Disorder suspected	
		Classical Galactosaemia - Gal-1-PUT deficiency	Galactokinase deficiency
	Main presentation	Liver dysfunction / failure to thrive / ecoli sepsis etc, (±cataracts)	Cataracts
Test	Galactosaemia Screen	Qualitative assay <ul style="list-style-type: none"> • First line if NOT transfused in preceding 4 months 	
	Galactose-1-phosphate (Gal-1-P)	<ul style="list-style-type: none"> • Monitoring treatment • If transfused – can be used as a screening test 	
	Galactose-1-phosphate uridyl transferase (GALT)	Quantitative assay <ul style="list-style-type: none"> • Confirmation • Identification of carriers and variants • Parents of transfused babies to exclude carrier status. 	

		<ul style="list-style-type: none"> Babies with reduced activity /delayed fluorescence on galactosaemia screens <p>Babies with raised Phe and Tyr levels on newborn screening</p>	
	Galactitol - urine	<ul style="list-style-type: none"> If transfused – can be used as a screening test 	<ul style="list-style-type: none"> First line screen Monitoring treatment
	Galactokinase		<p>Quantitative assay</p> <ul style="list-style-type: none"> Confirmation

The following assays are accredited to ISO 15189:2012

Qualitative galactosemia screen by fluorometric method (GAL)

First line test for classical galactosaemia, provided patient has not had a red blood cell transfusion in the previous 4 months. Sample types: Lithium Heparin whole blood (preferred), dried bloodspots also acceptable.

This test cannot exclude other forms of galactosaemia, carrier status or low activity variants. **TAT 4 days (urgently on request)**

Quantitative Galactose-1-phosphate uridyl transferase enzyme activity by tandem mass spectrometry (GALT)

This test can be used to confirm a diagnosis of classical galactosaemia, identify carriers and variant forms (e.g., Duarte galactosaemia), provided the patient has not received any red cell transfusions within last 4 months.

TAT 28 days

Galactokinase enzyme activity by tandem mass spectrometry (GALK)

This quantitative test can be used to confirm a diagnosis of galactokinase deficiency. Please phone our laboratory before sending as assay must be performed within 5 days of collection due to stability issues (Lithium heparin whole blood, no more than 24 hours at room temperature, ideally ship with an ice pack). **TAT 28 days**

Quantitative urine galactitol by GCMS (GTL)

Galactitol levels are raised in both classical galactosaemia and galactokinase deficiency. Can be helpful in excluding galactosaemia in patients who have had a blood transfusion. **TAT 28 days (urgently on request)**

Galactose-1-Phosphate by tandem mass spectrometry (Gal-1-P).

(This is available for routine analysis; UKAS accreditation is pending).

Most commonly used for monitoring classical galactosaemia: The acceptable level for a galactosaemic on a galactose free diet is less than 0.60 $\mu\text{mol/g Hb}$. **TAT 28 days**

If classical galactosaemia is suspected in a child that has had a red blood cell transfusion in the preceding 4 months, we would recommend requesting galactitol or Gal-1-P (from the child) or quantitative GALT activity from both the child's biological parents.

Should you have any queries relating to this service these can be directed to Principal Clinical Scientist Maryam Khan Maryam.Khan@nbt.nhs.uk

Galactosaemia Investigations Request form can be found - www.nbt.nhs.uk/severn-pathology/pathology-services/clinical-biochemistry/metabolic-biochemistry

9. Work Referred Away

The department may refer specimens to other specialist centres to provide a comprehensive diagnostic service. Most medical laboratories to which samples are referred are UKAS accredited. The performance of referral laboratories is routinely monitored. Where work has been done in other centres, this is made clear on our laboratory report.

10. Information Governance

We have high standards for the handling of patient and personal information. We maintain the necessary safeguards for, and appropriate use of, patient and personal information.

Privacy Policy and Data Protection

How we handle your information

Local Health Services such as hospitals, like ourselves, and GP Practices will record and keep your information to ensure you receive the best possible care. These records include:

- Your name, date of birth, NHS Number and contact details
- Information about your appointments and clinical visits
- Reports and notes about your health, treatment, and care
- Relevant information about people who care for you, such as next of kin or other healthcare professionals.

This information provides essential reference for Health Professionals who you see, in all parts of the NHS. It also enables us to investigate any issues, complaints, or legal claims.

All NHS Staff have a legal duty to keep your information confidential and secure and records are held securely and in the strictest confidence.

For further information on this please see <https://www.nbt.nhs.uk/about-us/information-governance/privacy-policy-data-protection>.