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| **Department of Clinical Biochemistry**Core Clinical Services DirectorateOur Ref: BS/CB/QM/COMU/146 |  Newborn Screening and Metabolic BiochemistryPathology Sciences BuildingNorth Bristol NHS TrustSouthmead HospitalSouthmead RoadBristolBS10 5NBTel: 0117 414 8425Email: [nbn-tr.ClinicalBiochemistryNBT@nhs.net](https://web.nhs.net/OWA/redir.aspx?C=qsTeG-fSK0CzUONimbPKfGiAZyNQgNFI-WSeUG204HNM5p8hM8LtF2hEmHCvIpDD5oasPAR5vQs.&URL=mailto%3anbn-tr.ClinicalBiochemistryNBT%40nhs.net)**Website:** [**www.severnpathology.com**](http://www.severnpathology.com/) |

Date 25th July 2022

Dear Colleague

**Galactosaemia Investigations**

We would like to take this opportunity to let you know we are in a position to offer a full range of assays for the investigations and monitoring of Galactosaemia within the Metabolic Biochemistry Department here at Southmead Hospital in Bristol.

The following assays are accredited to ISO 15189:2012

* **Qualitative galactosemia screen by fluorimetric method** (GAL)

Price £37.77

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-phoshate uridyl transferase enzyme activity by tandem mass spectrometry** (GALT)

Price £160.00

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)

Price £200.00

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)

Price £69.30

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).

Price £85.00

Most commonly used for monitoring classical galactosaemia: The acceptable level for a galactosaemic on a galactose free diet is less than 0.60 µmol/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.

An advantage of each of the tandem mass spectrometry methods is the smaller sample volume requirement. For each assay only 0.5ml of Li heparin whole blood (minimum volume) is required. Due to the smaller sample requirement and ability to run larger batches, we have been able to perform studies on sample stability and update our requirements accordingly. For example, galactokinase samples are now accepted later after collection than previously. See [www.nbt.nhs.uk/severn-pathology/requesting/test-information](http://www.nbt.nhs.uk/severn-pathology/requesting/test-information) for full details of sample requirements. Another advantage is none of the assays use enzymes as reagents, which reduces the associated risk of occasional supply/quality issues.

Attached is a comprehensive galactosaemia request form, this includes all available assays, sample requirements and contact numbers for the laboratory. Please use this for any referred samples. To set up NPEx requesting please contact our IT team winpathhelp@nbt.nhs.uk using ‘NPEx Galactosemia requests’ in the subject title.

The team here at the Metabolic Laboratory at Southmead Hospital are always available to offer telephone guidance for galactosaemia requesting or for advice regarding results, should it be required.

Should you have any queries relating to this service these can be directed to Principal Clinical Scientist Maryam Khan Maryam Khan@nbt.nhs.uk Alternatively, further information is available on our website: [www.severn-pathology.com](http://www.severn-pathology.com)

Yours sincerely



Mrs Nicola Crabbe

Lead Healthcare Scientist

**Galactosaemia Investigations**

Details of sample requirements can be found online <https://www.nbt.nhs.uk/severn-pathology/requesting/test-information> or by clicking the test names below.

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|  |  | **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**](https://www.nbt.nhs.uk/severn-pathology/requesting/test-information/galactosaemia-screen) | Qualitative assay * First line if NOT transfused in preceding 4 months
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| [**Galactose-1-phosphate (Gal-1-P)**](https://www.nbt.nhs.uk/severn-pathology/requesting/test-information/galactose-1-phosphate) | * Monitoring treatment
* If transfused – can be used as a screening test
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| [**Galactose-1-phosphate uridyl transferase (GALT)**](https://www.nbt.nhs.uk/severn-pathology/requesting/test-information/galactose-1-phosphate-uridyl-transferase-quantitative) | Quantitative assay * Confirmation
* Identification of carriers and variants
* Parents of transfused babies to exclude carrier status.
* Babies with reduced activity /delayed fluorescence on galactosaemia screens
* Babies with raised Phe and Tyr levels on newborn screening
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| [**Galactitol - urine**](https://www.nbt.nhs.uk/severn-pathology/requesting/test-information/galactitol) | * If transfused – can be used as a screening test
 | * First line screen
* Monitoring treatment
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| [**Galactokinase**](https://www.nbt.nhs.uk/severn-pathology/requesting/test-information/galactokinase) |   | Quantitative assay * Confirmation
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