

## NEWBORN SCREENING TEST REPERTOIRE

Department of Medical Biochemistry and Immunology  
University Hospital of Wales  
Heath Park  
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CF14 4XW

Newborn Screening section open Monday - Friday 08:45 to 17:15

For Laboratory general enquiries Tel: 029 21844032

Contact: Mr Luke Griffiths (Senior Biomedical Scientist) [Luke.Griffiths2@Wales.NHS.UK](mailto:Luke.Griffiths2@Wales.NHS.UK)

For clinical advice contact:

Professor Stuart J Moat (Consultant Clinical Scientist & Director - Wales Newborn Screening Laboratory)  
[Stuart.Moat@Wales.NHS.UK](mailto:Stuart.Moat@Wales.NHS.UK) Tel: 029 21843562

| Assay                                                           | Sample Requirement (ml)                                           | Sample handling / Transport          | Methodology                 | Reference Range                                                   | Turnaround Time (maximum working days) | EQA                                                                 |
|-----------------------------------------------------------------|-------------------------------------------------------------------|--------------------------------------|-----------------------------|-------------------------------------------------------------------|----------------------------------------|---------------------------------------------------------------------|
| Plasma Amino Acids                                              | 0.5ml lithium heparin plasma                                      | Send by first class post             | Ion Exchange Chromatography | Age related reference intervals & Interpretative report provided. | 10                                     | CDC Amino Acid Quality Assurance Program<br>CDC Proficiency Program |
| CSF Amino Acids                                                 | 0.5ml CSF (must be accompanied by a plasma sample)                | Send by first class post             | Ion Exchange Chromatography | Age related reference intervals & Interpretative report provided. | 10                                     | CDC Amino Acid Quality Assurance Program<br>CDC Proficiency Program |
| Urine Cystine Quantitation                                      | 5-10 ml aliquot of a random or 24 hour urine                      | Send by first class post             | Ion Exchange Chromatography | Reference intervals & Interpretative report provided.             | 5                                      | No EQA or alternative                                               |
| Urine Amino Acids (Quantitative)                                | 5 ml aliquot of a random or 24 hour urine collection              | Send by first class post             | Ion Exchange Chromatography | Age related reference intervals & Interpretative report provided. | 10                                     | CDC Amino Acid Quality Assurance Program<br>CDC Proficiency Program |
| Galactose-1-Phosphate Uridyl Transferase (Galactosaemia Screen) | 0.5ml lithium heparin blood                                       | Send whole blood by first class post |                             | Interpretative report provided.                                   | 10                                     | CDC Proficiency Program                                             |
| Congenital CMV Testing <sup>+</sup>                             | Laboratory staff will retrieve the Newborn Screening Guthrie card | N/A                                  | PCR                         | Interpretative report provided.                                   | 15                                     | -                                                                   |

<sup>+</sup> If testing for congenital CMV is required, please contact Professor Stuart Moat (details on page 1) prior to requesting the test. Test referred to Royal Free Hospital in London.

Prices on application