

## NEWBORN SCREENING TEST REPERTOIRE

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

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

For Laboratory general enquiries Tel: 029 21844032

Contact: Mr Chandra Sundas (Senior Biomedical Scientist) [Chandra.Sundas@Wales.nhs.uk](mailto:Chandra.Sundas@Wales.nhs.uk) Tel: 02921 843561

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 ( <i>maximum</i> 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  | UK NEQAS for Quantitative Amino Acids.<br>ERNDIM Quantitative amino acids scheme. |
| 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  | UK NEQAS for Quantitative Amino Acids.<br>ERNDIM Quantitative amino acids scheme. |
| 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.             | 10  | ERNDIM  |
| 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  | UK NEQAS for Quantitative Amino Acids.<br>ERNDIM Quantitative amino acids scheme. |
| 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.                                   | 6-8 weeks (includes time for sample retrieval and referral to external laboratory). | -   |

<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.