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Bwrdd Iechyd Prifysgol
Caerdydd a'r Fro
Cardiff and Vale
University Health Board

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 Luke Griffiths (Senior Biomedical Scientist) Luke.Griffiths@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 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. 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. 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	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	UK NEQAS for Quantitative Amino Acids. 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 ⁺	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).	-

⁺ 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.