

METABOLIC TEST REPERTOIRE

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

Metabolic section open Monday to Friday 08:45 to 17:15

For Laboratory general enquiries Tel: 029 21843560

Contact: Lesley Heirene (Senior Biomedical Scientist) Lesley.Heirene@wales.nhs.uk

For clinical advice contact:

Professor Stuart J Moat (Consultant Biochemist & Director - Wales Newborn Screening Laboratory)
Stuart.Moat@wales.nhs.uk Tel: 029 21843562

Claire Gallagher (Senior Clinical Scientist)
Claire.Gallagher2@wales.nhs.uk Tel: 02921 845448

Dr Thomas Lewis (Senior Clinical Scientist)
Thomas.Lewis5@wales.nhs.uk Tel: 02921 845448

Azza Osman (Consultant Clinical Pathology and Metabolic Medicine)
Azza.Osman@wales.nhs.uk Tel: 029 21840883

REQUESTING TESTS FOR ANALYSIS

Legible and fully completed request forms must accompany all samples, relevant information such as medications and date/time of sample collection are also provided. It is crucial that appropriate **CLINICAL DETAILS** are provided with all requests to ensure accurate clinical interpretation.

FOR URGENT REQUESTS PLEASE CONTACT THE LABORATORY TO DISCUSS WITH A CLINICAL SCIENTIST

Samples will be processed as soon as it is feasible to do so.

It may be possible to process small sample volumes but advisable to contact laboratory for advice.

QUALITY ASSURANCE

The Department participates in national and international external quality assurance schemes to monitor the accuracy and precision of its analyses.

The Metabolic/Special Chemistry Section participates in the following External Quality Assurance Schemes:

EQA Scheme	Assays covered
ERNDIM Special Assays in DBS	Acylcarnitines (free carnitine)
ERNDIM Acylcarnitines in DBS (qualitative)	Assessment of interpretation of acylcarnitine profile in light of clinical details
ERNDIM Special Assays in Serum	Non-esterified Fatty acids (NEFAs), 3-hydroxybutyrate (3OHB), Total homocysteine, Biotinidase, Methylmalonic acid (MMA) - plasma
ERNDIM Quantitative Organic Acids in Urine	Methylmalonic acid (MMA) - urine
ERNDIM Qualitative Organic Acids	Qualitative organic acids
ERNDIM Special Assays in Urine	Glycosaminoglycans (GAGs)
ERNDIM Diagnostic Proficiency Scheme	Test of laboratory's overall ability to apply the correct tests to a sample given clinical details and to correctly ascertain the diagnosis
ERNDIM Quantitative amino acids scheme	Amino acids
NEQAS	Total homocysteine, Sweat testing, vitamins A and E, Faecal Calprotectin, Faecal Elastase
UK NEQAS for Quantitative Amino Acids.	Amino acids
Willink MPS sample exchange scheme	Glycosaminoglycans (GAGs) – emphasis on qualitative assessment of GAGs results in clinical context

Metabolic tests:

Assay	Sample Requirement	Sample Handling/ Transport	Methodology	Reference Range	Turnaround Time (<i>maximum working days</i>)	Clinical Utility
Acylcarnitines	Capillary blood spotted onto a Guthrie card and dried at room temperature. Lithium heparin whole blood can be accepted and spotted out in the laboratory	Store at room temperature Send directly to laboratory via first class post. NB - ALWAYS treat Guthrie card as if it is a sample container and send a completed REQUEST FORM with it. DO NOT send to Newborn Screening	FIA-MSMS	Interpretative report provided	10	For the diagnosis of fatty acid oxidation disorders and some organic acidaemias
Amino Acids (Plasma)	0.5ml lithium heparin plasma	Samples should be stored frozen at -20°C Send by first class post	Ion Exchange Chromatography and LC-MS	Age related reference intervals & Interpretative report provided.	10	For the diagnosis of inherited disorders of amino acid metabolism and urea cycle defects.
Amino Acids (CSF)	0.5ml CSF (must be accompanied by a plasma sample)	Samples should be stored frozen at -20°C Send by first class post	Ion Exchange Chromatography	Age related reference intervals & Interpretative report provided.	10	To investigate primary amino acid disorders associated with neurological symptoms/seizures. NB-A paired plasma amino acid sample is essential.

Amino Acids (Urine) Qualitative	5 ml aliquot of a random or 24 hour urine collection	Samples should be stored frozen at -20°C Send by first class post	Ion Exchange Chromatography	Interpretative report provided.	10	Renal tubular disorders of amino acid transport.
Biotinidase	1ml plasma	Sample should be separated within 6 hours of collection Store at -20°C Send frozen on dry ice	Enzymatic	5.0-14.9 U/L	15	Biotinidase deficiency
Cystine (Urine) 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	Cystinuria investigation/monitoring
Glycosaminoglycans screen (GAG / creatinine ratio)	5ml fresh urine	Store at -20°C Send via first class post	Colourimetric	Age-related reference ranges (contact lab if you wish to discuss)	20	Screening test for mucopolysaccharide disorders
Homocysteine	0.5ml plasma EDTA	Separate within 30 mins of collection. Store -20°C and send first class post	CMIA	0-16 µmol/L	10	Homocystinuria/Cardiovascular
Methylmalonic acid (Plasma)	1ml plasma	Store at -20°C Send via first class post	LC MS/MS	0.10-0.42µmol/L	20	Vitamin B12 deficiency, NO abuse, diagnosis and monitoring patients with methylmalonic aciduria and cobalamin defects
Methylmalonic Acid (Urine)	3-5ml urine	Store at -20°C Send via first class post	LC MS/MS	0.8-3.4 mmol/mol creat	20	

Assay	Sample Requirement	Sample Handling / Transport	Methodology	Reference Range	Turnaround Time (maximum working days)	Clinical Utility
Non-esterified Fatty acids (NEFAs) 3-Hydroxybutyrate (3OHB)	1ml fluoride oxalate or EDTA plasma	Store at -20°C Send via first class post	Enzymatic	Interpretative report provided (blood glucose result at time of collection required)	20	Investigation of hypoglycaemia NB – a paired glucose result is essential for accurate interpretation
Organic Acids	Recommended sample volume 5mL urine Minimum sample volume 2.5mL	Store at -20°C Send via first class post	GCMS	Interpretative report provided	10	For investigation into diagnosis of inherited metabolic disorders
Sweat Test	Appointments by arrangement	-	Macroduct collection & Coulometric titration	Contact lab	72hrs following collection Urgent - available same day	Cystic fibrosis

Special Chemistry tests:

Assay	Sample Requirement	Sample Handling / Transport	Methodology	Reference Range	Turnaround Time (maximum working days)	EQA	Clinical Utility
Faecal Calprotectin	1-5g Faeces	Overnight room temp	ELISA	<50µg/g	10	U.K.NEQAS	Ulcerative colitis/Crohn's
Faecal Elastase	3g Faeces	Overnight room temp	ELISA	contact lab	10	U.K.NEQAS	Pancreatic exocrine dysfunction
Plasma Vitamins A and E	2ml Lithium heparin plasma	Protect from light within 24 hours of collection. Store plasma at -20°C. Send by first class post	LCMS	contact lab	10	U.K.NEQAS	Nutritional Assessment
Xanthochromia	200µl CSF	Centrifuge & protect supernatant from light, -20°C	Spectro-photometric scan	contact lab	1	U.K.NEQAS Sheffield	SAH

Please note that there is a surcharge for non-NHS patients as follows:

An additional charge of £5 for tests less than £20

An additional charge of 25% for those tests greater than £20

