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

## 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: Heather Wheatley (Senior Biomedical Scientist) [Heather.Wheatley@wales.nhs.uk](mailto:Heather.Wheatley@wales.nhs.uk)

For clinical advice contact:

Dr Stuart J Moat (Consultant Biochemist & Director - Wales Newborn Screening Laboratory)  
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Dr Duncan Cole (Consultant in Medical Biochemistry and Metabolic Medicine)  
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### FOR URGENT REQUESTS PLEASE CONTACT THE LABORATORY

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.

Please provide clinical details, particularly current patient medication, and collection times of sample on all request forms.

**Metabolic tests:**

Assay	Sample Requirement	Sample Handling/Transport	Methodology	Reference Range	Turnaround Time ( <i>maximum working days</i> )	EQA	Clinical Utility	NHS Charge # 2020/21
Acylcarnitines	Blood spot Guthrie card	Overnight Room temp	LC MS/MS	Interpretative report provided	10	CDC Atlanta and ERNDIM	Inborn errors of metabolism	£83.53
Biotinidase	1ml plasma	-20°C Sample should be separated within 6 hours of collection	Enzymatic	5.0-14.9 U/L	15	Sample exchange	Biotinidase deficiency	£49.02
Glycosaminoglycans (GAG Screen)	5ml fresh urine	-20°C	Colourimetric	contact lab	20	ERNDIM MPS and Willink	Mucopolysaccharide disorders	£36.84
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	U.K.NEQAS Sheffield and ERNDIM	Homocystinuria/ Cardiovascular	£26.59
Methylmalonic acid (Plasma)	1ml plasma	Overnight Room temp	LC MS/MS	0.10- 0.42µmol/L	20	UKNEQAS Holotranscobalamin	Vitamin B12 deficiency	£62.12
Methylmalonic Acid (Urine)	3-5ml urine	Overnight Room temp	LC MS/MS	0.7-3.4 mmol/mol creat	20	UKNEQAS Holotranscobalamin and ERNDIM	Vitamin B12 deficiency	£62.12

Assay	Sample Requirement	Sample Handling / Transport	Methodology	Reference Range	Turnaround Time (maximum working days)	EQA	Clinical Utility	NHS Charge # 2020/21
Non-esterified Fatty acids (NEFAs) 3-Hydroxybutyrate	1ml fluoride oxalate or EDTA plasma	-20°C	Enzymatic	Interpretative report provided (blood glucose result at time of collection required)	20	ERNDIM SAS	Investigation of hypoglycaemia	£17.97 £13.56
Organic Acids	3-5ml Urine	-20°C	GCMS	Interpretative report provided	10	ERNDIM Sheffield	Inborn errors of metabolism	£90.11
Very long chain fatty acids (VLCFA), including pristanic acid and phytanic acid	1ml plasma	Store frozen until dispatch	GCMS	C26: 0.33 – 1.39 µmol/L C26/C22: 0.000 – 0.030 C24/C22: 0.32 – 1.07 Pristanic acid and phytanic acid are also included, with age-related reference ranges. A full interpretative report is provided.	30	ERNDIM SAS	Investigation of peroxisomal disorders	£83.25
Sweat Test	Appointments by arrangement	-	Gibson and Cooke QPIT	Contact lab	Reports available same day	U.K.NEQAS Birmingham	Cystic fibrosis	£52.59

<b>Assay</b>	<b>Sample Requirement</b>	<b>Sample Handling / Transport</b>	<b>Methodology</b>	<b>Reference Range</b>	<b>Turnaround Time (maximum working days)</b>	<b>EQA</b>	<b>Clinical Utility</b>	<b>NHS Charge# 2020/21</b>
Urine Oxalate	20ml from 24hr acid collection	Overnight Room temp	enzymatic	contact Lab	10	WEQAS	Investigation of renal stones	£23.01
Urine Citrate	20ml from 24hr plain/acid collection	Overnight Room temp	enzymatic	contact Lab	10	WEQAS	Investigation of renal stones	£18.44
Urine Oxalate/Creatinine ratio	5ml random urine	Overnight Room temp	enzymatic	contact Lab	10	WEQAS	Investigation of renal stones	£24.45
Urine Citrate/Creatinine ratio	5ml random urine	Overnight Room temp	enzymatic	contact Lab	10	WEQAS	Investigation of renal stones	£19.89

## Special chemistry

Assay	Sample Requirement	Sample Handling / Transport	Methodology	Reference Range	Turnaround Time (maximum working days)	EQA	Clinical Utility	NHS Charge# 2020/21
Faecal Calprotectin	1-5g Faeces	Overnight room temp	Enzyme Immunoassay	<50µg/g	10	U.K.NEQAS	Ulcerative colitis/crohn's	£32.69
Faecal Elastase	3g Faeces	Overnight room temp	Elisa	contact lab	10	U.K.NEQAS	Pancreatic exocrine dysfunction	£33.71
Plasma Vitamins A and E	2ml plasma	protect from light, -20°C	HPLC	contact lab	10	U.K.NEQAS	Nutritional Assessment	£25.19
Xanthochromia	200µl CSF	protect from light, -20°C	Spectrophotometric scan	contact lab	1	U.K.NEQAS Sheffield	SAH	£28.91

**# 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**