University College London Hospitals
NHS Foundation Trust

**National Hospital for Neurology and Neurosurgery**
Basement, Albany Wing
Queen Square
London
WC1N 3BG

Laboratory direct line: *020 344 83818*
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# NEUROMETABOLIC UNIT

UKAS accredited medical laboratory number 8341
Supraregional Assay Service Laboratory

(Please check https://www.ukas.com/search-accredited-organisations/for confirmation of current status)

## A guide to services for users

8341

**Postal address for all samples:**

**Neurometabolic Unit (Box 105)**
**Basement, Albany Wing**
**The National Hospital for Neurology and Neurosurgery**
**Queen Square**
**London**
**WC1N 3BG**
**United Kingdom**
**Neurometabolic Unit Website:**
**https://www.uclh.nhs.uk/OurServices/ServiceA-Z/Neuro/NMU/Pages/Home.aspx**

## Background

The Neurometabolic Unit exists as an autonomous clinical laboratory within UCLH Queen Square Division to continue the provision and development of the highly specialised testing needed to support the diagnosis and treatment of neurological patients both within the UCLH Trust and nationally. On-going research programmes supplement and enhance the service provision. The laboratory has close collaborations with other specialist laboratories at NHNN and in the Queen Square area. Several members of staff have honorary or joint appointments with the Dept. of Chemical Pathology at Great Ormond Street Hospital (GOSH). The laboratory also hosts the diagnostic mitochondrial biochemistry laboratories for the NHS Rare Mitochondrial Disorders Service in London (http://mitochondrialdisease.nhs.uk/nhs-mitochondrial-services/london/)

# KEY STAFF AND CONTACT DETAILS

**Director: Consultant Clinical Scientist**

| | | |
|---|---|---|
| Prof Simon Heales | Tel(1): | 020 344 83844 (NHNN) |
| | Tel(2): | 020 7813 8321 (GOSH) |
| | Email(1): | simon.heales@gosh.nhs.uk |
| | Email(2): | simon.heales@nhs.net |
| | Email(3): | s.heales@ucl.ac.uk |

**Deputy Director: Principal Clinical Scientist**

| | | |
|---|---|---|
| Dr. Amanda Lam | Tel(1): | 020 344 83844 / 020 344 83818 (NHNN) |
| | Tel(2): | 020 7813 8495 (GOSH) |
| | Email(1): | amanda.lam@nhs.net |
| | Email(2): | amanda.lam@gosh.nhs.uk |
| | Email(3) | amanda.lam@ucl.ac.uk |

**Laboratory Manager: BMS 3**

| | | |
|---|---|---|
| Nana Ghansah | Tel: | 020 344 83844 / 020 344 83819 |
| GOSH honorary contract | Email: | nana.ghansah@nhs.net |

**Queen Square Division Laboratories Quality Manager**

| | | |
|---|---|---|
| Dr Vaneesha S Gibbons | Tel: | 020 344 84254 |
| | Email: | vaneeshagibbons@nhs.net |

**Senior Biomedical Scientist**

| | | |
|---|---|---|
| Sehrish Haidri | Tel: | 020 344 84716 / 020 344 83818 |
| GOSH honorary contract | Email: | sehrish.haidri@nhs.net |
| Dr. Simon Pope | Tel: | 020 3448 3835/ 020 344 83818 |
| | Email: | simonpope@nhs.net |

**Specialist Biomedical Scientist**

| | | |
|---|---|---|
| Caroline Bhairo | Tel: | 020 3448 3818 |
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| Amalia Lilian Stratiotis | Tel: | 020 3448 3818 |
| | Email: | amalia.stratiotis@nhs.net |

## Access to services

Located in the basement floor of the Albany Wing of The National Hospital for Neurology and Neurosurgery, the Unit is open from 9 a.m to 5:30 p.m Monday to Friday.

For laboratory enquiries please e-mail
**uclh.enquiry.neurometabolic@nhs.net**
For specimen reception enquiries please ring 020 344 83198
For urgent interpretative advice, please contact 020 344 83818 to be directed to the Duty Scientist. Otherwise, use laboratory e-mail for non-urgent enquiries.

An *out of hours* Clinical Scientist advisory service is available via Great Ormond Street Chemical Pathology Department. Please contact the main switchboard on **020 7405 9200.**

## User feedback contacts

For any user feedback, be they comments, requests, complaints or compliments please contact Prof Simon Heales, Dr. Amanda Lam, or Mr. Nana Ghansah in the first instance either verbally, via letter or email. If the complaint cannot be resolved locally, it will be escalated in accordance in with the Trust Complaints Procedure
https://www.uclh.nhs.uk/PandV/Helpandsupport/Commentssuggestionsandcomplaints/Pages/Home.aspx

We prepare an electronic user survey annually. Please help us improve our services by completing this survey. Please follow the link below:
https://www.surveymonkey.co.uk/r/WT5KFL6

## Service Level Agreements

The Neurometabolic Unit has a documented quality commitment to our laboratory users. Please use this in place of a service level agreement. See Appendix A for details of this commitment.

# REFERRING A SAMPLE TO OUR LABORATORY

## Sample collection and transport to us

The laboratory only accepts samples that have been collected following informed patient consent. It is the responsibility of our users to ensure that this is the case and it is assumed that samples sent to the laboratory have been collected with informed consent.

Special conditions apply to the collection and storage of samples for many of our tests and failure to comply will almost certainly invalidate results. To avoid this risk, especially with CSF and muscle, it is essential that these instructions are followed. *If in doubt please contact the laboratory for advice **before** taking samples.* Sample collection protocols are included in all our referral forms, available on our website.

**Referring laboratories are reminded that there are no facilities on site to receive and store samples outside of our operating hours shown above.**

## Test requesting and sample requirements

All non-UCLH specimens received by the laboratory should be accompanied with a written request for testing. All requests and specimens should contain 3 points of identification to unequivocally trace the laboratory result to the correct patient. Specific sample requirements are included with the details of the test.

Many of the specialist tests are time consuming, expensive to perform and interpretation is not possible without clinical information. Tests will not normally be reported with an interpretative comment unless adequate clinical information is provided on the request form or by personal contact.

## Data protection

The laboratory follows the ULCH Trust data protection policy.

## Add on tests

Add on tests are not usually recommended for the majority of our analytes due to their unstable nature. However, because of the often invasive collection procedure, we will consider adding on a test if the quality of the result will not be compromised. Please contact the laboratory to discuss appropriateness and time limits for requesting additional tests. Verbal add on requests **must** always be followed up by a written request.

## Turnaround times and Urgent requests

Target turnaround times are given for each test. Every effort is made to adhere to these times. If a result is required sooner than our quoted turnaround time, please contact the duty scientist or the named scientist for the test to discuss the urgency of the request. We will endeavour to expedite results in special circumstances.

## EQA performance

Where an EQA scheme is indicated for a particular test, it is implicit that performance in that scheme is satisfactory and NO communications regarding consistent poor performances have been received from the scheme organisers. Where an EQA is not available for tests offered by the laboratory the department has implemented alternative approaches including internal quality control procedures; these are documented as part of the quality management system.

## Research only Tests (R)

Tests marked with this symbol **R** have not been included in our scope for UKAS accreditation and therefore are not UKAS accredited tests. These tests are performed on an adhoc basis with no quoted turnaround time provided. These test have not been validated to ISO 15189 standards and are offered on a research basis only. Please discuss with the laboratory if you wish to send samples for one of our research tests. *Research samples will generally not be accepted unless the laboratory is contacted prior to sending a sample.*

## Tests performed in Neuroimmunology

Users often send samples for the Neuroimmunology laboratory and the Neurometabolic unit in the same package and we forward them internally; however, we are unable to answer queries regarding those tests. Please direct your enquiries to Neuroimmunology on 020 344 83814

# NEUROMETABOLIC UNIT REPERTOIRE

**R** denotes research test.

| Amino acids (plasma or CSF, quantitative) | |
|---|---|
| **Sample type** | **Heparinised or EDTA plasma, serum or CSF. Urine is only accepted for cystine quantitation (see separate entry)** |
| **Minimum volume** | **0.5 ml** |
| **Special requirements** | **Separate and freeze without delay. Courier frozen.** |
| **Notes** | **Please provide clinical details. It is strongly recommended that CSF is accompanied by a paired plasma sample. Diagnosis of glycine encephalopathy (NKH) is dependent on CSF/plasma glycine ratio from paired samples. Includes plasma total homocysteine for all samples. Please contact if plasma homocystine analysis is required. Contact: Nana Ghansah** |
| **Reference range** | **See report for full ranges. Ranges are derived in house.** |
| **Turnaround time** | **95% inside 4 working days** |
| **QA scheme** | **ERNDIM amino acids** |

| Angiotensin Converting Enzyme (CSF ACE) | |
|---|---|
| Sample type | **CSF** |
| Minimum volume | **0.5 ml** |
| Special requirements | **Stable for 4 days at ambient temperature, 10 days at 4°C, 6 months at -20°C.** Please send in LP4 or similar small tube not 30ml pots please as racking the large variety of tubes we receive is difficult. |
| Notes | **Specific tandem mass spectrometry method developed for the low levels found in CSF. This method has not been clinically validated as a marker of neuroinflammation, nor been clinically validated for the diagnosis and / or monitoring of neurosarcoidsis. Activity may be increased if blood brain barrier impairment leads to passage of serum proteins (including serum ACE) or blood contamination.** |
| Reference range | **0 – 1.20 µmol/min/L**<br>**Derived in house** |
| Turnaround time | **95% inside 25 working days** |
| QA scheme | **Alternative approach** |

| Aromatic amino acid decarboxylase (AAA decarboxylase; AADC) **R** | |
|---|---|
| **Sample type** | **Lithium heparin or EDTA plasma.** |
| **Minimum volume** | **250ul of plasma although 0.5-1ml preferred.** |
| **Special requirements** | **The blood sample should be centrifuged and the plasma should be frozen as soon as possible, stored at -70°C or below and transported to the NMU frozen in dry ice.** |
| **Notes** | **Aromatic amino acid decarboxylase (AAA decarboxylase; AADC) is a key enzyme in the synthesis of the neurotransmitters, dopamine and serotonin. It is a pyridoxal-5-phosphate (PLP) dependent enzyme and plasma/CSF PLP and CSF monoamine metabolites are useful adjuncts to this test if a disorder of neurotransmitter metabolism is suspected.<br>Contact Prof. Heales or Simon Pope** |
| **Reference range** | **Age dependent reference intervals derived in house provided with report.** |
| **Turnaround time** | **Contact laboratory to arrange investigation** |
| **QA scheme** | **None available** |

| Carnitine (free, total and acylcarnitine profile) | |
|---|---|
| **Sample type** | **Heparinised/EDTA plasma or serum** |
| **Minimum volume** | **0.5 ml** |
| **Special requirements** | **Separate serum / plasma within 3 hours of venepuncture and store frozen. Send by first class post or courier frozen if possible. Clinical details essential. Contact Sehrish Haidri** |
| **Notes** | **Measured by tandem mass spectrometry.<br>Laboratory report of Plasma Carnitine profile includes numerical concentration of total carnitine, free carnitine, acyl carnitine and an interpretative comment on the acylcarnitine profile if indicated.** |
| **Reference range** | **Total: 26 – 62 μmol/L. Free: 22 – 50 μmol/L. Acyl: 4 – 12 μmol/L. Derived in house** |
| **Turnaround time** | **95% inside 7 working days** |
| **QA scheme** | **Free carnitine and selected acylcarnitines - ERNDIM special assays in plasma and ERNDIM acylcarnitines in serum.** |

| Urine Cystine, Ornithine, Arginine and Lysine | |
|---|---|
| **Sample type** | **Plain or acidified urine. Random or 24 hr collection** |
| **Minimum volume** | **5 ml** |
| **Special requirements** | **Please indicate 24 hr volume in order to report excretion rates. If sending an aliquot, ensure thorough mixing beforehand. Specimens should be frozen as soon as possible.** |
| **Notes** | **This assay is not for urine amino acids *per se* but is tailored to the quantitation of cystine, ornithine, lysine and arginine for the diagnosis and monitoring of Cystinuria. Samples with urine pH of >8 will not be analysed due to bacterial contamination from specimen deterioration.** |
| **Reference range** | **Multiple ranges per creatinine and per 24hrs on report. Derived in house** |
| **Turnaround time** | **95% inside 4 working days** |
| **QA scheme** | **Participation in an inter-laboratory comparison scheme** |

| Homocysteine (total) | |
|---|---|
| **Sample type** | **Heparinised/EDTA plasma or serum** |
| **Minimum volume** | **0.2 ml** |
| **Special requirements** | **Ensure prompt (within 30 minutes) separation of plasma from cells. Separated serum/plasma is stable at room temp or 4°C for 3 days. Store frozen for longer time periods.** |
| **Notes** | **Plasma homocysteine is a sensitive functional indicator of vitamin B12 (and folate) status and a better primary screening test than serum B12 assay. Urine is not appropriate for screening. This analyte tends to increase with age but this is most likely secondary to declining B12 absorption and/or renal function.<br>Measured by tandem mass spectrometry.** |
| **Reference range** | **5 – 12 μmol/L<br>In house derived** |
| **Turnaround time** | **95% inside 5 working days** |
| **QA scheme** | **WEQAS homocysteine and ERNDIM special assays (plasma)** |

| Methymalonic acid / Methylmalonate (plasma) | |
|---|---|
| Sample type | **Heparinised/EDTA plasma or serum** |
| Minimum volume | **0.5 ml** |
| Special requirements | **Separate promptly and store frozen. Plasma may be sent by first class post** |
| Notes | **This tandem mass spectrometry assay is specifically for plasma and is not suitable for assay of urine excretion. Clinical details essential**<br>IN PARTICULAR PLEASE INDICATE IF PATIENT IS A KNOWN METHYLMALONIC ACIDURIA (including Cobalamin (CBl) metabolism disorders) |
| Reference range | **In the context of a raised total homocysteine and the absence of renal impairment:**<br>**< 0.29 μmol/l are considered not indicative of B12 deficiency**<br>**0.29-0.70 μmol/l suggests B12 deficiency**<br>**> 0.70 μmol/l consistent with overt B12 deficiency**<br>**Supported by in house data** |
| Turnaround time | **95% inside 7 working days** |
| QA scheme | **ERNDIM special assays (plasma) and Instand scheme** |

| 5-Methyltetrahydrofolate (CSF) | |
|---|---|
| **Sample type** | **Lumbar CSF** |
| **Minimum volume** | **0.5 ml** |
| **Special requirements** | **Please contact the laboratory prior to sampling for instructions on how to proceed with this investigation. The assay uses the second 0.5 ml of sequential lumbar CSF samples. Must be frozen immediately, stored at -70$^{O}$C or below and transported on dry ice. Clinical details are essential. Please see CSF neurotransmitter request form.** |
| **Notes** | **Clinical details essential. Interpretation on report. Please provide the paired peripheral folate level (if available) with the request to aid interpretation. For clinical advice, please contact Prof. S. Heales** |
| **Reference range** | **Age dependant reference intervals on report. Derived in house and published here: Awopetu, F. 2004. MSc Clinical Biochemistry Thesis, UCL.** |

| | |
|---|---|
| **Turnaround time** | **95% inside 30 working days** |
| **QA scheme** | **Alternative approach** |

| Mitochondrial Respiratory Chain Enzymes (and mitochondrial studies) | |
|---|---|
| Sample type | **50 – 100 mg skeletal muscle or 10-20 mg liver frozen. For fibroblast studies please contact unit** |
| Special requirements | **Must be frozen in dry ice or liquid nitrogen immediately, stored and transported in dry ice. Clinical details are essential. Please see the MRCE referral form.** |
| Notes | **Muscle and fibroblasts free of charge for NHS patients under NHS Highly Specialised Service (HSS) agreement – contact lab for private patient charges. The assays include Complex I, II-III, and IV assessment normalised to citrate synthase activity. Blue Native Gel analysis is undertaken if indicated to assess assembly* and Complex V disorders. Interpretation provided on report. Contact Dr Amanda Lam for protocol and advice.**<br>***Assessment of assembly is not UKAS accredited and is a research only test.**<br>**Associated tests include CSF 5-methyltetrahydrofolate and Coenzyme Q10 – see ubiquinone.** |
| Reference range | **Tissue dependant. Derived in house and published here: Heales SJR, Hargreaves IP, Olpin SE, et al, (1996), *J Inher Metab Dis* 19 (Supplement 1): P151** |
| Turnaround time | **95% inside 40 working days for Complex I, II-III and IV. A further 80 working days if complex V analysis is required.** |
| QA scheme | **Participation in an inter-laboratory comparison scheme** |

| Monoamine metabolites (CSF HVA and 5-HIAA) | |
|---|---|
| Sample type | **lumbar CSF** |
| Minimum volume | **0.5 ml** |
| Special requirements | **Please contact the laboratory prior to sampling for instructions on how to proceed with this investigation (prepared tubes from Neurometabolic Unit required). The assay uses the first 0.5 ml of sequential lumbar CSF samples. Freeze immediately, store at -70$^{O}$C and transport frozen. Clinical details essential. Please see CSF neurotransmitter request form.** |
| Notes | **The primary catabolic pathway for dopamine and serotonin produces Homovanillic Acid (HVA) and 5-Hydroxyindoleacetic acid (5-HIAA) respectively. CSF HVA and 5-HIAA measurements provide both diagnostic information and treatment monitoring for patients with inborn errors of neurotransmitter metabolism, movement disorders in both children and adults and other neurological disorders affecting the dopamine and serotonin pathways. A paired plasma for Prolactin can be helpful. Patients should ideally be off L-dopa medication for 7 days prior to lumbar puncture. If this is contraindicated, please contact to discuss alternative investigations. Contact Prof Heales for clinical advice** |
| Reference range | **Age dependent reference intervals. Derived in house And published here: Hyland K, Surtees RAH, Heales SJR et al. Cerebrospinal fluid concentrations of Pterins and metabolites of Serotonin and Dopamine in a pediatric reference population. *Pediatric Research.* 1993.** 34**: 10-14.** |
| Turnaround time | **95% inside 30 working days** |
| QA scheme | **ERNDIM Scheme** |

| Neopterin |
|---|
| **See pterins** |

| Neurotransmitters (CSF) |
|---|
| **See monoamine metabolites** |

| Phenylalanine / Tyrosine (blood spot) | |
|---|---|
| Sample type | **Blood spots on Guthrie card** |
| Minimum volume | **2 spots** |
| Special requirements | **Free falling drops of blood to be used, do not overlay spots, allow to dry before placing card in protective glassine envelope then into postal envelope. Send by first class post.** |
| Notes | **This service is used to monitor known cases of phenylketonuria (PKU) and tyrosinaemia only. Although we use the same methodology we are NOT a screening laboratory.** |
| Reference range | **n/a – monitoring levels defined by Charles Dent metabolic team at NHNN.** |
| Turnaround time | **95% inside 1 working day** |
| QA scheme | **CDC Newborn Screening; ERNDIM special assays in dried blood spot** |

| Phenylalanine Loading test |
|---|
| **Please see protocol at the end of the handbook** |

| Pterins (CSF) | |
|---|---|
| **Sample type** | **Lumbar CSF** |
| **Minimum volume** | **1 ml** |
| **Special requirements** | **Please contact the laboratory prior to sampling for instructions on how to proceed with this investigation (prepared tubes from Neurometabolic Unit required). The assay uses the third ml of sequential lumbar CSF samples, which should contain 1mg DTE + 1mg DETAPAC. Must be frozen immediately, stored and transported at -70$^{O}$C or below. Clinical details are essential. Please see CSF neurotransmitter request form.** |
| **Notes** | **BH4, BH2 and neopterin measurements provide diagnostic information regarding disorders and diseases affecting serotonin and dopamine metabolism within the central nervous system, particularly inborn errors of BH4 metabolism, for example atypical phenylketonuria and Dopa responsive dystonia. Neopterin is also a measure of immune response activation. Contact Prof. Heales for clinical advice** |
| **Reference range** | **Age dependent reference intervals. Derived in house And published here: Hyland K, Surtees RAH, Heales SJR et al. Cerebrospinal fluid concentrations of Pterins and metabolites of Serotonin and Dopamine in a pediatric reference population. *Pediatric Research.* 1993.** 34**: 10-14.** |
| **Turnaround time** | **95% inside 30 working days** |
| **QA scheme** | **Alternative approach** |

| Pyridoxal-5-phosphate (vitamin B6) in plasma | |
|---|---|
| **Sample type** | **EDTA plasma** |
| **Minimum volume** | **0.5 ml** |
| **Special requirements** | **Protect from light. Must be frozen immediately, stored and transported frozen with dry ice. Clinical details are essential** |
| **Notes** | **Pyridoxal-5-Phosphate (PLP) is the physiologically active form of vitamin B6 required for neurotransmitter synthesis and amino acid metabolism. Plasma and CSF amino acids are useful adjuncts to this test for the investigation of intractable seizures.Contact Prof. Heales** |
| **Reference range** | **Plasma 15 – 73 nmol/L<br>Literature values – Greiling H. et al Lehrbuch d. Klinischen Chemie und Pathobiochemie, Verlag Schattauer, 3 Auflg.., 1995, S. 458 supported by in house data.** |
| **Turnaround time** | **95% inside 10 working days** |
| **QA scheme** | **Instand** |

| Pyridoxal-5-phosphate (vitamin B6) in CSF | |
|---|---|
| Sample type | **CSF** |
| Minimum volume | **0.5 ml** |
| Special requirements | **Please contact the laboratory prior to sampling for instructions on how to proceed with this investigation (prepared tubes from Neurometabolic Unit required). The assay uses the second 0.5 ml of sequential lumbar CSF samples. Protect from light. Must be frozen immediately, stored and transported frozen with dry ice.. Clinical details are essential. Please see CSF neurotransmitter request form.** |
| Notes | **We strongly suggest a paired plasma is provided for measurement of peripheral levels to aid interpretation. Plasma and CSF amino acids are useful adjuncts to this test for the investigation of intractable seizures. Contact Prof. Heales** |
| Reference range | **Age related reference ranges are provided on the report. Derived in house and published here: Footitt E, Heales SJ, Mills PB, Allen GF, Oppenheim M, Clayton PT, J Inher Metab Dis. 2011 Apr;34(2):529-38** |
| Turnaround time | **95% inside 20 working days** |
| QA scheme | **Alternative approach** |

| Riboflavin (vitamin B2) **R** | |
|---|---|
| Sample type | **EDTA whole blood** |
| Minimum volume | **0.5ml** |
| Special requirements | **Store and transport whole blood frozen at -20°C and protected from light.** |
| Notes | **Dietary riboflavin is converted into the biologically active forms flavin adenine dinucleotide (FAD) and flavin mononucleotide (FMN). FAD and FMN act as cofactors for numerous redox enzymes. This sample may also be used for vitamin B1.**<br>**Contact Dr S Pope** |
| Reference range | **174 - 471 nmol/L (FAD) In house derived values.** |
| Turnaround time | **Contact laboratory to arrange investigation** |
| QA scheme | **Instand scheme** |

| Serotonin (blood) **R** | |
|---|---|
| Sample type | **EDTA whole blood (**tube should contain 5 mg ascorbic acid**)** |
| Minimum volume | **2 ml** |
| Special requirements | **Must be frozen immediately, stored and transported frozen on dry ice. Clinical details are essential** |
| Notes | **Serotonin is a major neurotransmitter. Quantification of serotonin in whole blood, the majority of which is contained in platelets, is useful in the assessment of serotonin metabolism in patients with suspected metabolic disorders affecting the availability of this neurotransmitter. This test has not been designed for the diagnosis or monitoring of carcinoid syndrome. Please contact the laboratory if an ascorbate containing EDTA tubes is required. Clinical details essential. Contact Prof. Simon Heales** |
| Reference range | **500 – 1600 nmol/L** |
| Turnaround time | **Contact laboratory to arrange investigation** |
| QA scheme | **None available** |

| Thiamine Pyrophosphate (vitamin B1) | |
|---|---|
| Sample type | **EDTA whole blood** |
| Minimum volume | **1ml** |
| Special requirements | **Stored frozen at -20°C as whole blood and transported frozen. Protect from light.** |
| Notes | **Dietary thiamine is converted into the biologically active form, thiamine pyrophosphate (TPP). TPP is an essential cofactor for numerous enzymes, including pyruvate dehydrogenase (PDH). Deficiency of TPP can lead to wet beriberi, dry beriberi, Wernicke's encephalopathy and Korsakoff's psychosis and can be observed following malnourishment and/ or malabsorption.<br>Contact Dr S Pope or Nana Ghansah** |
| Reference range | **67 – 265 nmol/L In house derived values.** |
| Turnaround time | **95% inside 15 working days** |
| QA scheme | **Instand** |

| Ubiquinone (muscle) | |
|---|---|
| Sample type | **10 – 20 mg frozen skeletal muscle** |
| Special requirements | **Must be frozen immediately, stored at -70°C and transport frozen on dry ice. Clinical details are essential. Please see muscle ubiquinone request form** |
| Notes | **Coenzyme $Q_{10}$ ($CoQ_{10}$) is an essential component of the mitochondrial respiratory chain, as well as being a powerful cellular antioxidant.**<br>**Contact Dr Amanda Lam** |
| Reference range | **140 – 580 pmol/mg protein** |
| Turnaround time | **95% inside 40 working days. Please note the turn around time of this test is measured after the completion of the mitochondrial respiratory chain enzymes (if also requested).**<br>**In house derived values** |
| QA scheme | **Alternative approach** |

| Ubiquinone (white cells) **R** | |
|---|---|
| Sample type | **EDTA blood** |
| Minimum volume | **5 ml** |
| Special requirements | **Keep at room temperature, must be received in NMU within 24 hours of sampling to allow for processing mononuclear cell pellet before storage. Please ensure sample arrive before 12pm on Fridays.**<br>Clinical details essential. |
| Notes | **Coenzyme $Q_{10}$ ($CoQ_{10}$) is an essential component of the mitochondrial respiratory chain, as well as being a powerful cellular antioxidant. This assay has not been designed for the monitoring or Coenzyme $Q_{10}$ supplementation.**<br>**Contact Dr Amanda Lam** |
| Reference range | **37 - 133 pmol/mg**<br>**In house derived values published here: Duncan AJ, Heales SJ, Mills K, et al 2005, *Clin Chem* 51: 2380-82** |
| Turnaround time | **Please contact the laboratory prior to sending samples to discuss a turnaround time.** |
| QA scheme | **None available** |

| Vitamin A (retinol) | |
|---|---|
| Sample type | **Serum or plasma** |
| Minimum volume | **2 ml** |
| Special requirements | **Separate and freeze serum or plasma at –20$^{O}$C and send to our laboratory frozen. Protect from light.** |
| Notes | **Vitamin A is a lipid soluble vitamin and an antioxidant. Absorption from the gut can be affected in states of fat malabsorption. This sample can also be used for vitamin E assay.**<br>**Contact Nana Ghansah** |
| Reference range | **1-6yr 0.7 – 1.50μmol/L**<br>**6-12yr 0.9 – 1.70μmol/L**<br>**12-19yr 0.9-2.50μmol/L**<br>**>19yr 1.05 – 2.80μmol/L**<br>**Aligned with paediatric ranges from Great Ormond St, verified by in house data.** |
| Turnaround time | **95% inside 15 working days** |
| QA scheme | **NEQAS vitamins** |

| Vitamin E (α-tocopherol) | |
|---|---|
| Sample type | **Serum or plasma** |
| Minimum volume | **2 ml** |
| Special requirements | **Separate and freeze serum or plasma at –20$^{O}$C. Send to our laboratory frozen on dry ice.** |
| Notes | **Vitamin E is a lipid soluble vitamin and an antioxidant. Absorption from the gut can be affected in states of fat malabsorption. α-tocopherol is bound to lipoproteins, so lipid status is essential when interpreting results.**<br>**This sample can also be used for vitamin A assay.**<br>**Contact Nana Ghansah** |
| Reference range | **11.5 – 46.4 μmol/L Literature values: Lehrbuch der klinischen Chemie und Pathobiochemie, H.Greiling, A.M. Gressner, 3. Auflg., Verlag Schattauer Stuttgart/New York, 1987. Supported by in house validation.** |
| Turnaround time | **95% inside 15 working days** |
| QA scheme | **NEQAS vitamins** |

## Vitamin B1

**See thiamine**

## Vitamin B2

**See riboflavin**

## Vitamin B6

**See pyridoxal phosphate**

# PHENYLALANINE LOADING TEST

Please note: This protocol is taken directly from the publication of Hyland *et al* (1997), *Neurology*; **48**(5): 1290. This protocol has not been validated by the Neurometabolic Unit. This test is not UKAS accredited.

## For the investigation of dopamine responsive dystonia.

**Procedure:**

- Patients are allowed a low-protein breakfast (cereal) approximately 2 hours before the phenylalanine load. No further food is permitted before the phenyalanine load.
- Prepare 100mg/kg body weight of L-phenylalanine. The published protocol made up 10g of phenylalanine suspended in 100 ml of lemonade (not diet, i.e. not containing aspartame). The appropriate volume should be given to the patient.
- Take 1-2 ml heparinised blood (baseline) before the phenylalanine load and arrange for plasma to be separated and frozen.
- Ensure that the phenylalanine solution is well mixed. Give phenylalanine at approximately 10 am ensuring that it is completely consumed.
- Take further lithium heparin blood samples as above at 1, 2, 4 and 6 hours post dose. Arrange for each sample to be separated and the plasma frozen after each time point. Do not keep samples until test is completed. Ensure the sample is fully labelled with three points of ID. The patient should not have food during the test.
- If the patient is currently on L-DOPA this does not interfere with the physiological response.

If plasma phenylalanine samples are to be sent to the Neurometabolic Unit, please contact the laboratory before carrying out this test especially if additional tests such as CSF Neurotransmitters are also required. Telephone +44 (0)203 448 3818

Please contact simon.heales@nhs.net if an interpretation is required

# Appendix A: Quality Commitment

Provided that we are sent samples and accompanying requests that are valid and adhere to all referral requirements stated in our User Handbook we make the following assurances to you:

1. We will inform you as quickly as possible if we believe that the results provided for any test/assay and clinical/interpretive service, are for any reason, unreliable.
2. We will inform you as soon as possible of any circumstances that adversely affect our turnaround times or the quality of services that we provide for your referred samples.
3. Wherever available, we are registered with an EQA scheme, or interlaboratory comparison programme, appropriate to the service provided.
4. We will inform you of any adverse EQA that result in persistent poor performance and/or if we were to be contacted by the scheme organisers.
5. Where no EQA scheme or inter-laboratory comparison programme is available, we have alternative mechanisms in place to provide objective evidence for determining the acceptability of test/assay results.
6. We will inform you of any changes to sample requirements (including, but not limited to, sample volume, sample collection and transport conditions) for the testing we perform for you.
7. We will inform you of any changes that could lead to results or their interpretation being significantly different for the tests we perform for you.
8. We will notify you of any changes to our Quality Management System that could adversely influence the quality of results that we provide.
9. We will notify you of any change of contact details. **Please view our website for up to date information.**

The above commitment applies to all referred tests and associated interpretations that we provide for you and includes all aspects that are pre-defined in any individual agreements.

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|---|---|---|
| Prof Simon Heales<br>Laboratory Director | Dr. Amanda Lam<br>Deputy Laboratory Director | Mr. Nana Ghansah<br>Laboratory Manager |

Issued by:

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