



CARDIFF PORPHYRIA SERVICE TEST REPERTOIRE 2019/2020

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

Porphyria Service Open Monday – Friday 09:00 to 17:00

For Laboratory enquiries: 029 21843565 (Jackie Woolf)
For Genetics enquiries: 029 21842811 (Sharon Whatley)

Email enquiries: CAV_porphyria@wales.nhs.uk

For clinical advice: 029 21846588 (Dr Mike Badminton, mike.badminton@wales.nhs.uk)

Website: www.cardiff-porphyria.org

N.B. Please refer to the Sample Advice page on our website for help in choosing the most appropriate samples to send in relation to the patients' clinical state.

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. PLEASE CONTACT THE LABORATORY FOR ADVICE. PLEASE PROVIDE CLINICAL DETAILS AND RESULTS OF OTHER RELEVANT TESTS WHEREVER POSSIBLE. PLEASE STATE SAMPLE COLLECTION DATE ON ALL REQUEST FORMS.

| Routine porphyrin tests: | | | | | | | |
|---------------------------------|--|--|---------------------------------|--|--|---|--------------------------------------|
| Assay | Sample requirements (ml) | Sample handling / Transport | Methodology | Reference range | Turnaround Time Maximum TAT quoted in working days* | Clinical Utility | NHS Charge* 2019/20 £ |
| Total urine porphyrin | Urine, preferably early morning sample (2ml) | Overnight at room temperature. Protected from light | Spectro-fluorimetry | <40nm/mmol creat | 10 | Suspected porphyria. Monitoring bullous porphyria treatment | 19.58 |
| Porphobilinogen (urine) | Urine, preferably early morning sample (3ml) | Overnight at room temperature. Protected from light | Ion exchange/spectroscopy | <1.5umol/mmol creat | 10 | Suspected acute porphyria. Monitoring known acute porphyria patients. | 39.91 |
| Aminolevulinic acid (urine) | Urine, preferably early morning sample (4ml) | Overnight at room temperature. Protected from light | Ion exchange/spectroscopy | <3.8umol/mmol creat (Adult) <5.2umol/mmol creat (Child) | 15 | Limited availability. Please contact laboratory. | 41.58 |
| HPLC (urine) | Urine, preferably early morning sample (2ml) | Overnight at room temperature. Protected from light | HPLC fluorimetric detection | N/A | 15 | Confirmation of diagnosis | 48.95 |
| Total faecal porphyrin | Stool (~5g) | Overnight at room temperature. Protected from light) | Solvent extraction/spectroscopy | <200nmol/g dry weight | 15 | Diagnosis following positive screening tests | 37.05 |
| HPLC (Faeces) | Stool (~5g) | Overnight at room temperature. Protected from light) | HPLC fluorimetric detection | N/A | 15 | Confirmation of diagnosis | 48.95 |

| Assay | Sample requirements (ml) | Sample handling / Transport | Methodology | Reference range | Turnaround Time (working days) | Clinical Utility | NHS Charge* 2019/20 £ |
|---------------------------------|---------------------------|---|---------------------|-----------------|--------------------------------|---|-----------------------|
| Plasma porphyrin screen | EDTA whole blood (5-10ml) | Overnight at room temperature. Protected from light) | Spectro-fluorimetry | Not increased | 10 | Suspected porphyria | 19.58 |
| Red blood cell porphyrin screen | EDTA whole blood (5-10ml) | Overnight at room temperature. Protected from light)) | Spectro-fluorimetry | Negative | 10 | Suspected cutaneous porphyria | 20.00 |
| Red blood cell total porphyrin | EDTA whole blood (5-10ml) | Overnight at room temperature. Protected from light)) | Spectro-fluorimetry | 0.4-1.7umol/l | 15 | Suspected cutaneous porphyria, monitoring patients with EPP | 33.26 |

*Please contact the laboratory to arrange urgent analyses - all tests performed outside normal working hours will incur an additional charge of £70

*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

EQA- All assays registered for RCPA-AACB Australia & EPNET EQA