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University Health Board

## **CARDIFF PORPHYRIA SERVICE TEST REPERTOIRE 2023/2024**

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 (Lucy Bentley)  
For Genetics enquiries: 029 21842811 (Michelle Wood)

Email enquiries: [CAV\\_porphyria@wales.nhs.uk](mailto:CAV_porphyria@wales.nhs.uk)

For clinical advice: 029 21846588 (Dr Danja Schulenburg-Brand)  
[Danja.Schulenburg-brand@wales.nhs.uk](mailto:Danja.Schulenburg-brand@wales.nhs.uk)

Website: [www.cardiff-porphyria.org](http://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.**

<b>Routine porphyrin tests:</b>							
<b>Assay</b>	<b>Sample requirements (ml)</b>	<b>Sample handling / Transport</b>	<b>Methodology</b>	<b>Reference range</b>	<b>Turnaround Time Maximum TAT quoted in working days*</b>	<b>Clinical Utility</b>	<b>NHS Charge* 2023/24 £</b>
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	23.14
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.	47.14
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)	10	Limited availability. Please contact laboratory.	49.13
HPLC (urine)	Urine, preferably early morning sample (2ml)	Overnight at room temperature. Protected from light	HPLC fluorimetric detection	N/A	10	Confirmation of diagnosis	57.82
Total faecal porphyrin	Stool (~5g)	Overnight at room temperature. Protected from light)	Solvent extraction/spectroscopy	<200nmol/g dry weight	10	Limited availability. Please contact laboratory.	43.78
HPLC (Faeces)	Stool (~5g)	Overnight at room temperature. Protected from light)	HPLC fluorimetric detection	N/A	10	Confirmation of diagnosis	57.82

<b>Assay</b>	<b>Sample requirements (ml)</b>	<b>Sample handling / Transport</b>	<b>Methodology</b>	<b>Reference range</b>	<b>Turnaround Time (working days)</b>	<b>Clinical Utility</b>	<b>NHS Charge* 2023/24 £</b>
Plasma porphyrin screen	EDTA whole blood (5-10ml)	Overnight at room temperature. Protected from light)	Spectro-fluorimetry	Not increased	10	Suspected porphyria	23.14
Red blood cell porphyrin screen	EDTA whole blood (5-10ml)	Overnight at room temperature. Protected from light))	Spectro-fluorimetry	Not increased	10	Suspected cutaneous porphyria	23.63
Red blood cell total porphyrin	EDTA whole blood (5-10ml)	Overnight at room temperature. Protected from light))	Spectro-fluorimetry	0.4-1.7umol/l	10	Suspected cutaneous porphyria, monitoring patients with EPP	39.28

\*Please contact the laboratory to arrange urgent analyses - all tests performed outside normal working hours will incur an additional charge of £81.93

\*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 & IPNET EQA