



GIG
CYMRU
NHS
WALES

Bwrdd Iechyd Prifysgol
Caerdydd a'r Fro
Cardiff and Vale
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

For clinical advice: 029 21846588 (Dr Danja Schulenburg-Brand)
Danja.Schulenburg-brand@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.

Assay	Sample requirements (ml)	Sample handling / Transport	Methodology	Reference range	Turnaround Time Maximum TAT quoted in working days*	Clinical Utility
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
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.
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.
HPLC (urine)	Urine, preferably early morning sample (2ml)	Overnight at room temperature. Protected from light	HPLC fluorimetric detection	N/A	10	Confirmation of diagnosis
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.
HPLC (Faeces)	Stool (~5g)	Overnight at room temperature. Protected from light)	HPLC fluorimetric detection	N/A	10	Confirmation of diagnosis

Assay	Sample requirements (ml)	Sample handling / Transport	Methodology	Reference range	Turnaround Time (working days)	Clinical Utility
Plasma porphyrin screen	EDTA whole blood (5-10ml)	Overnight at room temperature. Protected from light)	Spectro-fluorimetry	Not increased	10	Suspected porphyria
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
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

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

For current prices please contact the laboratory.

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