

Porphyria Studies

Synonyms

Porphrins, Porphobilinogen, PBG, ALA

Clinical Indication

Clinical Presentation	Specimens	Comments
Acute neurological attacks (suspected AIP, VP, HCP)	Urine Faeces Blood	To exclude a current attack urine alone is adequate.
Acute photosensitivity (suspected EPP)	Blood	Urine and faeces of no value.
Skin lesions (suspected PCT, VP, HCP or CEP)	Urine Faeces Blood	To monitor patients with PCT send early morning spot urine only in white top universal, protected from light.

Investigation of suspected porphyria. The incidence of porphyria is such that an average District should contain about 20 patients with overt porphyria. The rate of diagnosis is much less than this and may be due to failure to suspect porphyria as a possible diagnosis.

Part of Profile / See Also

Request Form

Combined Pathology manual Blood form or ICE request. Relevant clinical information MUST be given.

Availability / Frequency of Analysis

Referred Test: Analysed at the Clinical Biochemistry Department, Bedford Hospital [8212](#), if specific criteria met. All requests will be vetted by Consultant Biochemist to ensure correct samples based on clinical details are referred.

Turnaround Time

The routine turnaround time is 1-2 weeks but urgent requests can be arranged with prior discussion with the Consultant Biochemist.

Patient Preparation

Sample Requirements

Specimen Type

Early Morning Urine and/or Faeces (small random sample approx.10g)
 Blood (plasma and red cells).

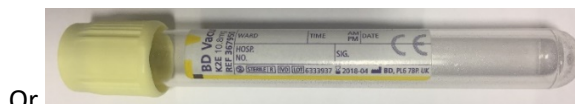
Volume

All samples must be protected from light.
 2ml (blood), 10 ml (urine), 10 g (faeces)

Container



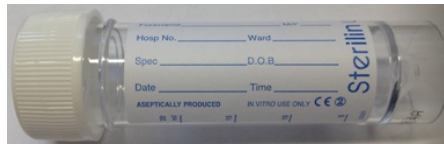
Pink/purple top (EDTA) tube



Lemon top (EDTA) tube



Green top (li-Heparin) tube



Urine and faeces should be collected into plain sterile containers.

All samples must be protected from light immediately after collection

Reference Range & Units

Provided within the interpretative report.

Interferences

Interpretation & Clinical

Decision Value (if applicable)

Acute neurological attacks: if urine screen normal and urine was collected during a suspected attack then no further tests are normally required.

An interpretive comment will be added to reports.

References

Test code

Lab Handling

Urine and/or Faeces must be stored at -20°C until sent to the referral laboratory.

The blood sample must be centrifuged and the plasma separated from the red cells; both the plasma and red cells must be stored at 4°C.

Urgent analysis can be arranged with prior approval from the Biochemist.

All samples must be protected from light prior to analysis/referral.

