



Cast 37 – Acute Porphyria

A 50-year-old woman presents to the Emergency Department with **severe abdominal pain**, nausea and vomiting for 24 hours. She reports **constipation**, dark-coloured urine, and anxiety. Examination reveals tachycardia and hypertension. Her abdomen is soft with minimal tenderness. Blood tests show **hyponatraemia** and a **high ferritin level**.

a) List 8 differential diagnoses for this presentation (8 marks)

Any **eight** of the following:

1. **Acute porphyria** (e.g. acute intermittent porphyria)
2. **Acute pancreatitis**
3. **Mesenteric ischaemia**
4. **Bowel obstruction**
5. **Lead poisoning / heavy metal toxicity**
6. **Diabetic ketoacidosis**
7. **Thyroid storm**
8. **Phaeochromocytoma crisis**
9. **Sepsis with abdominal source**
10. **Acute hepatitis**
11. **Drug or toxin ingestion**
12. **Intestinal pseudo-obstruction**

Examiner insight (why these score well)

Strong candidates usually recognise the “**abdominal pain + autonomic instability + hyponatraemia + minimal exam findings**” cluster, which is classic for **acute porphyria**, but they must also include:

- **Surgical catastrophes** → mesenteric ischaemia
- **Metabolic causes** → DKA
- **Toxicological causes** → heavy metals
- **Endocrine crises** → thyroid storm / pheochromocytoma



B) Clinical features suggestive of acute porphyria (6 marks)

Any **six** of:

- Severe **abdominal pain** out of proportion to examination findings
- **Autonomic dysfunction**: hypertension, tachycardia
- **Neuropsychiatric features**: anxiety, agitation, confusion, psychosis
- **Peripheral neuropathy**: limb weakness, paraesthesia
- **Dark or reddish urine**, often darkens on standing
- **Hyponatraemia** (often due to SIADH)
- **Constipation** rather than diarrhoea
- Seizures

C) List two (2) Precipitants of acute porphyria (2 marks)

Any **two** of:

- **Medications** (e.g. barbiturates, anticonvulsants, sulfonamides)
- Rifampicin, Ketorolac
- **Hormonal changes** (e.g. luteal phase of menstruation, pregnancy)
- OCP
- **Fasting or low-carbohydrate intake**
- Infection or physiological stress
- Alcohol use
- Organic solvents used in painting/cleaning

Anaesthetic precipitants

- Definite – barbiturates, phenytoin, sulphonamides, ropivacaine
- Possible – etomidate, lignocaine, chlordiazepoxide, diazepam, halothane, corticosteroids, ketamine, atracurium



D) List four (4) Initial investigations you would perform in a patient suspected of acute porphyria (4 marks)

Any **four** including bold of:

- **Urinary porphobilinogen (PBG)** – key diagnostic test (spot urine acceptable)
- **Urinary ALA (δ -aminolevulinic acid)**
- Full blood count
- **Urea, electrolytes (particularly sodium)**
- Liver function tests
- Serum osmolality / urine sodium if SIADH suspected
- Pregnancy test for female of child bearing age.
- ECG (electrolyte disturbance, autonomic effects)

Note: Imaging often normal and should not delay diagnosis if suspicion is high.

E) State six (6) key steps in Acute management in the ED (6 marks)

Any **six** of:

- **Cease precipitating drugs** (e.g. enzyme-inducing medications)
- **Early consultation** with toxicology / haematology / porphyria specialist
- **IV haem arginate** (definitive treatment for moderate–severe attacks)
- **High-carbohydrate therapy** (e.g. IV dextrose) if haem unavailable or mild attack
- Adequate **opioid analgesia** (avoid unsafe drugs)
- **Antiemetics** that are porphyria-safe
- Correct **electrolyte abnormalities**, particularly hyponatraemia (fluid restriction)
- Monitor for **respiratory failure and neuropathy** (ICU if severe)

Safe drugs

- Antacids – Na⁺ citrate
- Induction – propofol
- Inhalational agents – N₂O, halothane, isoflurane (use with caution)
- NMBD – sux, pancuronium and vecuronium
- Reversal – atropine, glycopyrulate, neostigmine
- Analgesia – opioids
- LA – bupivacaine
- Sedatives – midazolam, temazepam, lorazepam
- Antiemetics – droperidol
- Cardiovascular drugs – adrenaline, phenylephrine, salbutamol, Mg²⁺, beta-blockers, phentolamine