

Porphyria

- Rare, mostly inherited disorders of the haem pathway.
- Problems arise because of a build-up of haem precursors released from either the liver or bone marrow (the two sites of haem production).

Disclaimer:
Read the disclaimer at medimaps.co.uk/disclaimer

References:
1. Pulsetoday.co.uk

Acute porphyrias

- 1 in 20,000 have it (usually undiagnosed).
- Only 10 in 1 million have symptoms.
- Four subtypes:
- Acute intermittent porphyria (AIP)
- Variegate porphyria (VP)
- Hereditary coproporphyria (HCP)
- ALAD-deficient porphyria (ALAD-P)

Skin porphyrias

Symptoms

- Severe, poorly localised abdominal pain.
- Pain in back and legs.
- Nausea, vomiting and constipation.
- Agitation, insomnia, rarely psychosis
- Dark orange or red urine.
- Tachycardia and hypertension.
- Hyponatraemia.
- Neurological complications.

often misdiagnosed as

- Appendicitis
- Cholecystitis
- Endometriosis (premenstrual).
- Guillain-Barré syndrome (paralysis)
- Functional disorder or opiate seeking behaviour.

- Seizures
- Progressive motor neuropathy (leading to paralysis and respiratory failure in severe attacks).

Investigations

symptoms resolved

- Random light protected urine sample (plain container)
- Light-protected blood sample (full blood count tube)
- Small light-protected stool sample

Contact a specialist porphyria centre for advice

symptoms present

- Random light protected urine sample.
- For porphobilinogen and porphyrin levels.

Urine porphobilinogen excretion is always increased during an acute attack of porphyria while a normal result excludes an attack.

- Contact the National Acute Porphyria Service for advice about immediate management.
- Tel: 02920 747747

Management

- All patients (including those with a genetic diagnosis who have never had symptoms) should take steps to reduce their risk of an attack.
- In particular, all medication should be checked for safety in acute porphyria.
- Check safety on cardiff-porphyria.org.
- Most patients will benefit from referral to a specialist porphyria centre.