VERMAAK: EDUPATH

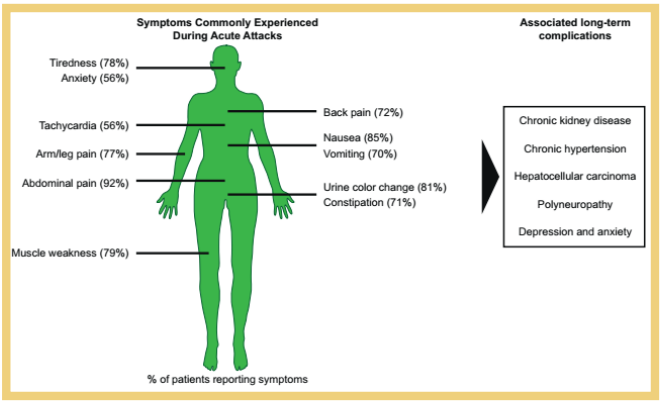
INVESTIGATION OF PORPHYRIA

The porphyrias arise from enzyme defects in the haem biosynthetic pathway. Accumulation of the precursors (ALA and PBG) lead to acute neurovisceral attacks, marked by severe abdominal pain and autonomic neuropathy, which may progress to a potentially fatal motor neuropathy. Accumulation of the porphyrins lead to photocutaneous sensitivity, which presents with skin fragility, blistering and erosions in sun-exposed areas. For further information refer to www.porphyria-professionals.uct.ac.za

Porphyria can be divided into the acute porphyrias and cutaneous porphyrias:

1. Acute porphyria

- Cause potentially life-threatening acute neurovisceral attacks
- May be precipitated by infection, alcohol, drugs, reduced calorie intake and stress
- Four disorders:
  - variegate porphyria (VP)
  - acute intermittent porphyria (AIP)
  - hereditary coproporphyria (HCP)
  - ALA dehydratase porphyria (ALAD)
- VP, AIP and HCP are autosomal dominantly inherited.



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2. Cutaneous porphyria

- Porphyria cutanea tarda (PCT; mainly acquired on a background of chronic liver disease) and congenital erythropoietic porphyria (CEP) result in skin fragility and blisters in sun-exposed areas. This may also be present in a proportion of VP and HCP
- Erythropoeietic protoporphyria (EPP) and X-linked EPP cause acute skin pain shortly after sun exposure, with lack of clinical signs.

The porphyrias most frequently encountered in South Africa are VP, AIP and PCT.

CHOOSING THE CORRECT TEST

Porphyria can be difficult to diagnose because of its diverse presentation, and overlap with many other conditions. Investigations should be requested according to clinical presentation:

1. Acute porphyric attack

- Random urine wrapped in foil
- Test code A3568 (acute porphyrias only: VP, AIP, ALAD and HCP)
- Includes: PBG/creatinine ratio, total porphyrins, fractionated porphyrins.

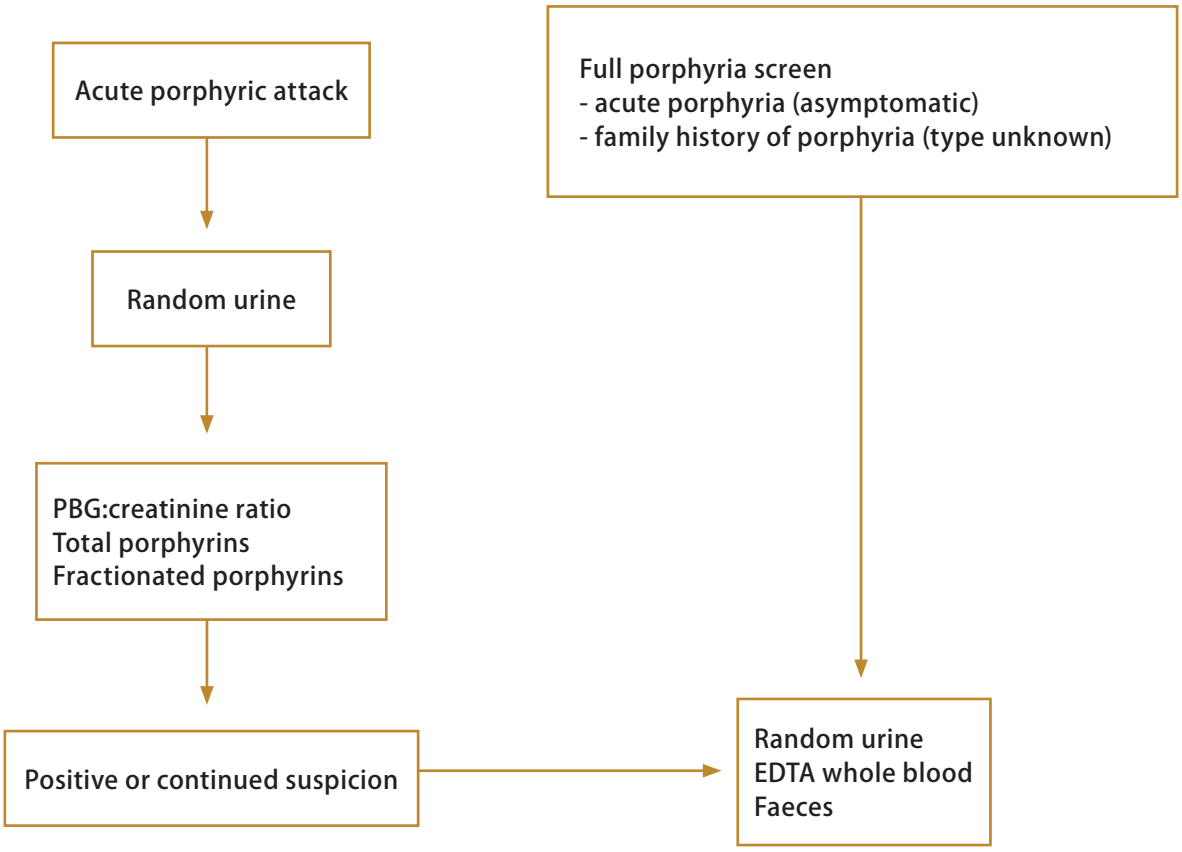
2. Full porphyria screen: asymptomatic acute porphyria, family history of porphyria (type unknown)

- Random urine **and** EDTA tube **and** stool sample all wrapped in foil
- Test code V1098 (all porphyrias)
- Includes: PBG/creatinine ratio, free porphyrins (urine, rbc and stool), fractionated porphyrins (urine and stool), plasma fluorescence scan.

3. Acute porphyria follow-up

- Random urine wrapped in foil
- Test code PBGDUB
- Includes: PBG/creatinine ratio.

Genetic testing for the R59W mutation is recommended if the biochemical analyses are suggestive of VP, or if there is a family history.



Reference: ACB (2017) 54(2)

Chemical Pathology, PathCare, 2023