**INDICATIONS FOR TESTING**

Clinical suspicion of porphyrias

- **Neurologic and abdominal symptoms**
  - Consider AIP, HCP, VP
  - Consider PCT, HCP, VP, CEP

- **Cutaneous photosensitivity**
  - Consider EPP/XLP

**ORDER**

- Erythrocyte Porphyrin (EP), Whole Blood
- Porphyrins, Fractionation and Quantitation, Urine
- Porphobilinogen (PBG), Urine
- Porphyrins, Fecal
- Aminolevulinic Acid (ALA), Urine

**INITIAL TESTING**

- Strong suspicion
  - Consider ADP (rare)
  - Aminolevulinic Acid (ALA), Urine
  - ADP probable
  - Confirmatory testing
    - Porphobilinogen (PBG) Deaminase, Erythrocyte
    - Variant analysis (PBGD gene)

- Negative
  - Repeat testing when active symptoms are present
  - Confirmatory testing
    - Aminolevulinic Acid Dehydratase (ALAD), Blood
    - Variant analysis (ALAD gene)

- Positive
  - Porphyria unlikely if symptomatic
  - Porphyrins, Fecal
  - Positve*:
    - ADP
    - HCP
    - VP
  - Positve*:
    - Porphyrin, Total, Plasma or Serum

**Conclusive testing**

- **Pseudoporphyria, immunobullous disease, or connective tissue disease possible**
  - Skin biopsy for direct immunofluorescence and serum for indirect immunofluorescence
  - Connective tissue disease workup

**Abbreviations**

- ADP: Aminolevulinic acid dehydratase-deficiency porphyria
- AIP: Acute intermittent porphyria
- CEP: Congenital erythropoietic porphyria
- EPP: Erythropoietic protoporphyria
- HCP: Hereditary coproporphyria
- PCT: Porphyria cutanea tarda
- VP: Variegate porphyria
- XLP: X-linked protoporphyria

*Result pattern, as interpreted by a medical expert, distinguishes porphyria type.
*If asymptomatic, consider repeating testing when symptoms are present.