**Porphyrias Testing**

**INDICATIONS FOR TESTING**

- Clinical suspicion of porphyrias

**Cutaneous photosensitivity**

- Neurologic and abdominal symptoms
- Consider AIP, HCP, VP
- Consider HCP or VP

**Cutaneous photosensitivity AND Neurologic and abdominal symptoms**

- Consider PCT, HCP, VP, CEP
- Consider EPP/XLP

**ORDER**

- Erythrocyte Porphyrin (EP), Whole Blood
- Porphyrins, Fractionation and Quantitation, Urine
- Porphyrins, Total, Plasma or Serum
- Porphyrins, Fecal

**Initial Testing**

- Strong suspicion
  - Consider ADP (rare)
  - Aminolevulinic Acid (ALA), Urine
  - ADP unlikely if asymptomatic

- Negative
  - Porphyrin unlikely if asymptomatic

- Positive
  - ORDER Porphyrins, Fecal
  - Negative
  - ORDER Aminolevulinic Acid Deaminase, Erythrocyte, Variant analysis (PBGD gene)
  - Negative
  - AIP
  - Positive
  - Confirmatory testing
    - Porphobilinogen (PBG) Deaminase, Erythrocyte
    - Variant analysis (PBGD gene)
  - Positive
  - Repeat testing when active symptoms are present

- Other testing
  - Porphyrins, Total, Plasma or Serum (use to monitor disease)
  - Porphyrins, fecal
  - Variant analysis (UROD gene)

- Other testing
  - Porphyrins, Fecal
  - May consider
  - Erythrocyte Porphyrin (EP), Whole Blood
  - Variant analysis (UROS gene)

- Pseudoporphryia, immunobullous disease, or connective tissue disease possible

**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.