Porphyrias Testing

INDICATIONS FOR TESTING
Clinical suspicion of porphyrias

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

- Cutaneous photosensitivity
  - AND
  - Neurologic and abdominal symptoms
  - Consider HCP or VP

- Cutaneous photosensitivity
  - Blistering
  - Burning, itching, redness
  - Consider PCT, HCP, VP, CEP

ORDER
- Erythrocyte Porphyrin (PBG), Urine

ORDER
- Porphyrins and Porphobilinogen (PBG), Urine

ORDER
- Porphyrins, Fractionation and Quantitation, Urine

ORDER
- Erythrocyte Porphyrin (EP), Whole Blood

INITIAL TESTING

- Strong suspicion
- Negative
- Positive

Consider AIP, HCP, VP

- Positive
- Porphyrin unlikely if symptomatic*

ORDER
- Aminolevulinic Acid (ALA), Urine

- Negative
- Repeat testing when active symptoms are present

- Positive
- AIP

Conversatory testing
- Porphobilinogen (PBG)
- Deaminase, Erythrocyte
- Variant analysis (PBGD gene)

ORDER
- Porphyrins, Total, Plasma or Serum

- Negative
- Positive*
- Porphyrin unlikely

ORDER
- Erythrocyte Porphyrin (EP), Urine

- Positive*

ORDER
- Aminolevulinic Acid Dehydratase (ALAD), Blood
- Variant analysis (ALAD gene)

ORDER
- Porphyrins, Total, Plasma or Serum

- Positive*
- Porphyrin unlikely

ORDER
- Variate analysis (UROD gene)

ORDER
- Variate analysis (PPOX gene)

ORDER
- Variate analysis (CPOX gene)

ORDER
- Variate analysis (FECH gene, ALAS2 gene)

*Result pattern, as interpreted by a medical expert, distinguishes porphyria type.

If asymptomatic, consider repeating testing when symptoms are present.