Porphyria (Acute) Testing Algorithm*

**Symptoms:**
- Neurovisceral attacks (abdominal pain, neuropathy, psychiatric symptoms)
- Tachycardia and hypertension

**Possible acute porphyria:**
- Acute intermittent porphyria (AIP)
- Variegate porphyria (VP)*
- Hereditary coproporphyria (HCP)*
- Aminolevulinic acid dehydratase deficiency porphyria (ADP)

**To differentiate ADP from tyrosinemia type I and heavy metal intoxication order:**
- ALAD / Aminolevulinic Acid Dehydratase (ALAD), Whole Blood*
- OAUC / Organic Acids Screen, Urine
- HMCRU / Heavy Metal/Creatinine Ratio, with Reflex, Urine OR
- HMDB / Heavy Metals Screen with Demographics, Blood

**Decreased ALAD activity**

- Confirms ADP

**Normal ALAD activity**

- Excludes ADP

**Isolated ALA increase**

- PQNRU / Porphyrins, Quantitative, Random, Urine
- Includes porphobilinogen
- PBALP / Porphobilinogen and Aminolevulinic Acid, Plasma*

**Increased porphobilinogen, possible increases in uroporphyrin, coproporphyrin, and/or ALA**

**Perform:**
- PBGD_/ Porphobilinogen Deaminase (PBGD), Whole Blood*
- FQPPS / Porphyrins, Feces

**Increased coproporphyrin III/I ratio (<10) and protoporphyrin**

- AIP – Family studies may be warranted
- HMBSZ / HMBS Gene, Full Gene Analysis, Varies

**Increased coproporphyrin III/I ratio (>10) and coproporphyrin III**

- VP – Family studies may be warranted
- PPOXZ / PPOX Gene, Full Gene Analysis, Varies

**Increased PBGD activity and fecal porphyrin profile**

- HCP – Family studies may be warranted
- CPOXZ / CPOX Gene, Full Gene Analysis, Varies
- Excludes VP and HCP
- AIP not excluded*

**Normal results - were samples collected during an acute episode?**

- YES
  - Excludes acute porphyrinas
  - Retest during acute episode
  - NO

**Possible acute porphyria:**
- Acute intermittent porphyria (AIP)
- Variegate porphyria (VP)*
- Hereditary coproporphyria (HCP)*
- Aminolevulinic acid dehydratase deficiency porphyria (ADP)

**Normal results - were samples collected during an acute episode?**

- YES
  - Excludes acute porphyrinas
  - Retest during acute episode
- NO

- A. 80% of patients with VP have cutaneous symptoms
- B. 20% of patients with HCP have cutaneous symptoms
- C. Specimens collected during symptomatic period will be most informative
- D. Plasma specimen for those unable to give urine
- E. ALAD test is not useful for lead intoxication cases
- F. 5% of AIP patients have normal PBGD activity in erythrocytes
- G. Specimens collected during asymptomatic period will be most informative.

*Interpretive report provided for all tests in this algorithm