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)

**Decreased ALAD activity**
- Excludes ADP
- Confirms AIP

**Normal ALAD activity**
- Isolated ALA increase
- Decreased PBGD activity
- Increased coproporphyrin III/I ratio (<10) and coproporphyrin III
- Increased coproporphyrin III/I ratio (>10) and coproporphyrin III
- Normal PBGD activity and fecal porphyrin profile

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

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

**Retest during acute episode**
- Increased porphobilinogen, possible increases in uroporphyrin, coproporphyrin, and/or ALA

**QONRU / Porphyrins, Quantitative, Random Urine**
- ALAUR / Aminolevulinic Acid (ALA), Urine

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

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

**HCP – Family studies may be warranted**
- CPOXZ / CPOX Gene, Full Gene Analysis

**Normal results – was urine collected during an acute episode?**
- YES: Excludes acute porphyras
- NO: Re却est during acute episode

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

**Increased coproporphyrin III/I ratio (<10) and protoporphyrin**
- AIP – Family studies may be warranted
- HMBSZ / HMBS Gene, Full Gene Analysis

**Increased coproporphyrin III/I ratio (>10) and coproporphyrin III**
- VP – Family studies may be warranted
- PPOXZ / PPOX Gene, Full Gene Analysis

**Increased coproporphyrin III/I ratio (>10) and coproporphyrin III**
- HCP – Family studies may be warranted
- CPOXZ / CPOX Gene, Full Gene Analysis

**Normal PBGD activity and fecal porphyrin profile**
- Excludes VP and HCP
- AIP not excluded*

**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.** ALAD test is not useful for lead intoxication cases
**e.** 5% of AIP patients have normal PBGD activity in erythrocytes
**f.** Specimens collected during asymptomatic period will be most informative.

*Interpretive report provided for all tests in this algorithm