Porphyria (Acute) Testing Algorithm*

**Decreased ALAD activity**
- Confirms ADP

**Normal ALAD activity**
- Excludes ADP

**Isolated ALA increase**
- PQNRU / Porphyrins, Quantitative, Random Urine
- ALAUR / Aminolevulinic Acid (ALA), Urine

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

**Increased coproporphyrin III/I ratio (<10)**
- 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

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

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

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

**Normal results—was urine collected during an acute episode?**
- YES
  - Excludes acute porphyras
- NO
  - Retest during acute episode

**To differentiate ADP from tyrosinemia type I and heavy metal intoxication order:**
- ALAD / Aminolevulinic Acid Dehydratase (ALAD), Whole Blood
- OAU / Organic Acids Screen, Urine
- HMSRU / Heavy Metals Screen, Random Urine or HMSBR / Heavy Metals Screen with Demographics, Blood

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

**Notes:**
- 80% of patients with VP have cutaneous symptoms
- 20% of patients with HCP have cutaneous symptoms
- Specimens collected during symptomatic period will be most informative
- ALAD test is not useful for lead intoxication cases
- 5% of AIP patients have normal PBGD activity in erythrocytes
- Specimens collected during asymptomatic period will be most informative

* Interpretive report provided for all tests in this algorithm