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)\(^a\)
- Hereditary coproporphyria (HCP)\(^b\)
- Aminolevulinic acid dehydratase deficiency porphyria (ADP)

**Decreased ALAD activity**
- Confirms ADP

**Normal ALAD activity**
- Excludes ADP

**Isolated ALA increase**
- PONRU / Porphyrins, Quantitative, Random Urine\(^c\)
- ALAUR / Aminolevulinic Acid (ALA), Urine\(^c\)

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

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

**Normal PBGD activity and fecal porphyrin profile**
- Excludes VP and HCP
- AIP not excluded\(^e\)

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

**Decreased PBGD activity**
- 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
- Retest during acute episode

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

**Perform:**
- PBGD / Porphobilinogen Deaminase (PBGD), Whole Blood\(^d\)
- FOOPPS / Porphyrins, Feces

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\(^*\) Interpretive report provided for all tests in this algorithm

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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.