Porphyria (Acute) Testing Algorithm*

Decreased ALAD activity

Normal ALAD activity

Isolated ALA increase

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

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

PQNRU / Porphyrins, Quantitative, Random Urine\(^c\)
- ALAUR / Aminolevulinic Acid (ALA), Urine\(^c\)

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

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

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

Decreased PBGD activity

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

\(^a\) 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.

Retest during acute episode

Normal results—was urine collected during an acute episode?

YES

Excludes acute porphyras

NO

Excludes VP and HCP

AIP not excluded\(^*\)

* Interpretive report provided for all tests in this algorithm