Diagnosing Acute Intermittent Porphyria: a Guide for Clinicians

Because signs and symptoms of acute intermittent porphyria (AIP) mimic other, more common diseases, diagnosis of AIP is challenging. AIP is a rare inherited disease caused by a deficiency of the enzyme porphobilinogen (PBG) deaminase in the heme biosynthetic pathway. Untreated attacks can result in neurologic damage or even death; therefore, early diagnosis and treatment of AIP are critical.

**PATIENT PRESENTATION**

- **Abdominal pain most common symptom**
  - Present in ≥ 85% of patients
  - Neuropathic in origin
  - Usually severe, unremitting, and diffuse

- **Other common acute symptoms**
  - Gastrointestinal
    - Vomiting
    - Constipation
    - Diarrhea
  - Urinary
    - Dark or reddish urine
  - Neurologic
    - Pain in the extremities, back, chest, neck, or head
    - Paresis
    - Respiratory paralysis
    - Mental symptoms
    - Convulsions
  - Cardiovascular
    - Tachycardia
    - Systemic arterial hypertension

- **Family history of acute intermittent porphyria (AIP)**

**PATIENT HISTORY**

**ACTION**

**PATIENT CHARACTERISTICS**

- Gender (acute attacks are 4 to 5 times more common in women)
- Luteal phase of menstrual cycle
- Age of patient (acute attacks most common in their 30s)
- and/or

**POSSIBLE PRECIPITATING FACTORS**

- Various drugs†
- Endogenous hormones
- Crash dieting
- Alcohol use
- Illicit drugs
- Smoking
- Stress

**PBG† urine test**

Should be done at or near the time of symptoms

Available through major clinical laboratory testing companies

**Index of Suspicion**

Enzymatic and DNA testing

For further confirmatory testing

*Based on several series of patients with symptomatic AIP.
†When evaluation does not support another cause, remember that atypical presentations can occur.
‡Not all symptoms in porphyric patients are due to porphyria—porphyric patients are not immune to other conditions.
¶In approximately one third of cases, family history is absent due to disease latency.
†PBG=porphobilinogen.

Lists of drugs thought to precipitate an AIP attack can be obtained through various publications and the American Porphyria Foundation website.

This guide is provided by Recordati Rare Diseases for educational purposes only and is not meant to substitute for the independent medical judgment of a healthcare provider relative to diagnostic and treatment options for a specific patient’s medical condition.