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

PATIENT CHARACTERISTICS

including

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

including

Various drugs†

Endogenous hormones

Crash dieting

Alcohol use

Illicit drugs

Smoking

Stress

Index of Suspicion

ACTION

PBG¶ urine test

Should be done at or near the time of symptoms

Available through major clinical laboratory testing companies

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.

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