Diagnosing Acute Intermittent Porphyria (AIP)

Diagnosis of AIP is challenging because signs and symptoms mimic other, more common conditions.[1] AIP is a rare inherited disease caused by a partial deficiency of the enzyme porphobilinogen (PBG) deaminase in the heme biosynthetic pathway.[1]

Accuracy and speed are critical in the diagnosis of an acute porphyric attack.[1]

Patient presentation

Abdominal pain most common symptom[1]

- Present in >85% of patients
- Neuropathic in origin
- Usually severe, unremitting, and diffuse

Other common acute symptoms†[1]

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‡[2]

Patient history

Patient characteristics

- Gender (acute attacks are 4 to 5 times more common in women)[2]
- Luteal phase of menstrual cycle[1]
- Age of patient (acute attacks most common in their 30s)[2]

Possible precipitating factors

- Use of alcohol or illicit drugs[1,2]
- Endogenous hormones[1]
- Crash dieting[1]
- Smoking[1,2]
- Emotional and/or physical stress[1,2]

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Action

PBG urine test

- Screening tests to measure the levels of the porphyrin precursor PBG in urine are essential to confirm a diagnosis of acute porphyria[3]
- Acute attacks are always accompanied by increased production and excretion of PBG in AIP[3,4]
- It is essential that before arranging for a PBG urine test, the physician consult with the laboratory to ensure that the test is available and also know the recommended procedures as to how and when to collect the urine sample[4]

Genetic testing

- AIP is metabolic disorder[3,4]
- For confirmatory molecular genetic testing, physicians should consult their provincial laboratory services for more information

† Not all symptoms in porphyric patients are due to porphyria – porphyric patients are not immune to other conditions.[2]
‡ In approximately one third of cases, family history is absent due to disease latency.[2]

References:
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2. Thadani H, Deacon A, Peters T. Diagnosis and management of porphyria. *BMJ.* 2000;320(7250);1647-1651.
3. National Organization for Rare Disorders. Rare Disease Database: Acute Intermittent Porphyria. Accessed November 21, 2019 at https://rarediseases.org/rare-diseases/acute-intermittent-porphyria/
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