IS IT ACUTE HEPATIC PORPHYRIA (AHP)?

DETECT

Severe, diffuse abdominal pain[1,2]

+

1 or more of the following signs and symptoms

AUTONOMIC Nervous System[1,2]	CENTRAL Nervous System[1-3]	PERIPHERAL Nervous System[1,2]
Nausea/vomiting Constipation Tachycardia Systemic arterial hypertension	Seizures Anxiety Mental status changes	Limb weakness or pain Peripheral neuropathy

CUTANEOUS[1]	OTHER Common AHP Symptoms[1,4]
Skin lesions on sun-exposed areas (Cutaneous symptoms primarily occur in HCP and VP.)	Hyponatremia Dark, reddish urine

>90% of patients with AHP* report abdominal pain during attacks (mimics an acute abdomen but without specialized localization)[1,2,5]

*There are 4 AHP types. About 80% of cases are acute intermittent porphyria (AIP), followed by variegate porphyria (VP), hereditary coproporphyria (HCP), and the extremely rare ALAD-deficiency porphyria (ADP).[1,6,7]

SUSPECT

Nonspecific symptoms can lead to misdiagnoses[1,8-10]

- ✗ **Irritable bowel syndrome**
- ✗ **Acute abdomen**
- ✗ **Endometriosis**
- ✗ **Fibromyalgia**
- ✗ **Psychiatric disorders**

- ✓ **AHP**

TEST

Random (spot) urine tests can be used to help diagnose[4]

- ✓ **PBG**† (porphobilinogen)
- ✓ **ALA**† (aminolevulinic acid)
- ✓ **Porphyrins**‡

Genetic testing can help confirm a biochemical test, help identify the specific mutation that a patient has, or help diagnose a patient outside an attack.[1,2,4§]

†PBG is highly specific to help diagnose AHP, while testing ALA can be helpful for differential diagnosis of ADP.[1]

‡It's not recommended to use urine porphyrins alone, as they can be elevated for several reasons.[1]

§Penetrance in AHP is low, so people with a gene mutation for AHP may not develop symptoms.[4]

Acute hepatic porphyria (AHP)

Devastating to miss

When the signs and symptoms make you suspect AHP, consider ordering all of the following random (spot) urine tests[4]

- **PBG**[11] CPT Code: 84110*
- **ALA**[11] CPT Code: 82135*
- **Porphyrins**[11] CPT Code: 84120*

Substantial elevation of urinary PBG is a hallmark indicator of 3 types of AHP: **acute intermittent porphyria (AIP)**, **variegate porphyria (VP)**, and **hereditary coproporphyria (HCP)**.[1,4] The majority of cases (~80%) of AHP are AIP.

*Depending on instruments and methodology used, some labs may use different CPT codes.

AHP is a rare genetic condition that can be devastating

AHP is characterized by acute, potentially life-threatening attacks. Patients with chronically elevated levels of ALA and/or PBG contribute to the development of long-term chronic complications, such as hypertension, hepatocellular carcinoma, and chronic kidney disease.[12,13]

Common symptoms

The cardinal symptom is severe, diffuse abdominal pain. Other common symptoms may include nausea and vomiting, limb weakness or pain, anxiety, and confusion.[1,2]

AHP most often occurs in women of childbearing age.[6] The major signs and symptoms are due to effects on the autonomic, central, and peripheral nervous systems.

Burden of delayed diagnosis

AHP is often misdiagnosed due to the nonspecific nature of symptoms, and patients may cycle from specialist to specialist.[2]

Delayed diagnosis or misdiagnosis of an AHP attack may lead to more severe attack symptoms and poorer patient outcomes, including hospitalizations and unnecessary surgeries.[1,10,13-16]

The pathophysiology of AHP

AHP is caused by an enzyme deficiency in the heme biosynthesis pathway in the liver. Disease triggers can induce ALAS1, the key regulator in this pathway, leading to the overproduction of neurotoxins ALA and PBG, which are factors associated with AHP attacks and other disease manifestations.[1,4]

The optimal time to test spot urine is during or shortly after an attack when ALA and PBG levels have spiked because levels may fall when symptoms resolve.[1]

CPT=Current Procedural Terminology.

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