### IS IT ACUTE HEPATIC PORPHYRIA (AHP)?

**Signs and symptoms of AHP** include \(^1\-^3\):

#### SEVERE, DIFFUSE ABDOMINAL PAIN

1 OR MORE OF THE FOLLOWING

<table>
<thead>
<tr>
<th>PERIPHERAL Nervous System</th>
<th>CENTRAL Nervous System</th>
<th>AUTONOMIC Nervous System</th>
<th>CUTANEOUS†</th>
</tr>
</thead>
<tbody>
<tr>
<td>• Limb weakness or pain</td>
<td>• Anxiety</td>
<td>• Nausea</td>
<td>• Skin lesions on sun-exposed areas</td>
</tr>
</tbody>
</table>

92% of patients with AHP report abdominal pain (mimics an acute abdomen but without specific localization)\(^3\-^5\).

*There are 4 AHP subtypes. About 80% of cases are acute intermittent porphyria (AIP), followed by hereditary coproporphyria (HCP), variegate porphyria (VP), and the extremely rare ALA dehydratase-deficiency porphyria (ADP).\(^1\-^4\)
†Cutaneous symptoms occur only in HCP and VP.\(^1\-^3\)

### Nonspecific symptoms can lead to misdiagnoses

- Irritable bowel syndrome
- Inflammatory bowel disease
- Endometriosis
- Fibromyalgia
- Psychiatric disorder
- AHP

### Confirm suspicion by running simple spot urine tests\(^1\-^3\)

- **PBG** (porphobilinogen)\(^2\)
- **ALA** (delta-aminolevulinic acid)\(^4\)
- **Porphyrens**

Urinary porphyrins is a nonspecific test and should not be used alone to diagnose AHP\(^5\).

\(^1\)PBG and ALA are porphyrin precursors that occur naturally in the heme biosynthesis pathway in the liver but reach neurotoxic levels in patients with symptomatic AHP.\(^1\-^2\)

\(^2\)Porphyrin analyses may differentiate the specific AHP.\(^1\)
A family of rare, genetic diseases

AHP features acute, potentially life-threatening exacerbations and, for some patients, chronic, debilitating symptoms. It may inflict years of suffering and impaired quality of life. \(^1\sim\)\(^5\)

AHP is driven by one of several enzyme defects in the heme biosynthesis pathway in the liver. These defects induce compensatory overexpression of ALA synthase 1 (ALAS1), resulting in neurotoxic accumulations of ALA and PBG and leading to disease manifestations. \(^1\),\(^3\)

The neurotoxic burden of ALA and PBG

ALA and PBG are normal precursors of porphyrin synthesis, but they are also neurotoxic in high concentrations. \(^1\)

ALA is believed to be the primary neurotoxin responsible for the triad of chronic symptoms, acute exacerbations, and long-term disease complications. Although less neurotoxic, PBG is highly specific as a diagnostic marker for AHP. \(^2\),\(^3\)

ALA and PBG should be tested along with porphyrins to confirm an AHP diagnosis. Normal urine PBG in symptomatic patients excludes the 3 most common subtypes of AHP as the cause of symptoms. Because ALA and PBG are most likely to be elevated during symptomatic periods, the timing of testing is important. \(^1\),\(^2\),\(^6\)

Incapacitating symptoms, mostly in females

Symptomatic disease most often occurs in women of childbearing age. The major signs and symptoms are due to effects on the nervous system. \(^2\),\(^3\)

While presenting symptoms vary, the cardinal symptom is severe, diffuse abdominal pain in up to 92% of patients. Other common symptoms may include nausea and vomiting, dark or reddish urine, confusion and anxiety, and limb pain or weakness. \(^3\),\(^5\)

In a cohort of patients with frequent exacerbations, up to 65% of patients also reported chronic symptoms and 46% reported daily symptoms. \(^5\)

Consequences of delayed diagnosis

AHP often escapes diagnosis because the symptoms overlap with those of numerous common conditions. \(^3\)

Without early diagnosis, patients may cycle from specialist to specialist and experience repeated hospitalizations, unnecessary surgeries, and long-term medical complications such as kidney disease and hypertension. \(^2\),\(^3\)

Patients with recurrent attacks may have been previously diagnosed with:

Viral gastroenteritis, irritable bowel syndrome, cholecystitis, appendicitis, hepatitis, endometriosis, depression, psychosis, stress, seizure disorder, appendicitis, Guillain-Barré syndrome, lead poisoning, or addiction withdrawal. \(^2\),\(^6\)–\(^8\)

When the signs and symptoms make you suspect AHP, order these spot urine tests to be sure \(^1\),\(^3\)

- **PBG** (porphobilinogen)  
  CPT Code: 84110

- **ALA** (delta-aminolevulinic acid)  
  CPT Code: 82135

- **Porphyrins** is a nonspecific test and should not be used alone to diagnose AHP  
  CPT Code: 84120

References:


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