

IS IT ACUTE HEPATIC PORPHYRIA (AHP)?

Severe, diffuse abdominal pain^{1,2}
+

1 or more of the following signs and symptoms

AUTONOMIC Nervous System^{1,2}

Nausea/vomiting
Constipation
Tachycardia
Systemic arterial hypertension

CENTRAL Nervous System¹⁻³

Seizures
Anxiety
Mental status changes

PERIPHERAL Nervous System^{1,2}

Limb weakness or pain
Peripheral neuropathy

CUTANEOUS¹

Skin lesions on sun-exposed areas (Cutaneous symptoms primarily occur in HCP and VP.)

OTHER Common AHP Symptoms^{1,4}

Hyponatremia
Dark, reddish urine

>90% of patients report abdominal pain during AHP* attacks (mimics acute abdomen but without specialized localization)^{1,2,5}

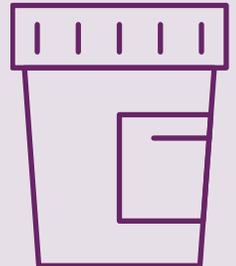
*There are 4 AHP types. About 80% of cases are acute intermittent porphyria (AIP), followed by variegate porphyria (VP), hereditary coproporphyria (HCP), and the extremely rare ALAD-deficiency porphyria (ADP).^{1,6,7}

Nonspecific symptoms can lead to misdiagnoses^{1,8-10}

- ✗ Irritable bowel syndrome
- ✗ Acute abdomen
- ✗ Endometriosis
- ✗ Fibromyalgia
- ✗ Psychiatric disorders
- ✓ AHP

Random (spot) urine tests are the primary method⁴

- ✓ PBG^{††} (porphobilinogen)
- ✓ ALA^{††} (aminolevulinic acid)
- ✓ Porphyrins[§]



Genetic testing can help determine the type of AHP a patient has and help with screening family members of patients with AHP^{1,2,4}

[†]ALA and PBG should be normalized to creatinine in order to adjust for differences in the degree of urinary concentration.

[‡]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

DETECT

SUSPECT

TEST

Acute hepatic porphyria (AHP) Making a diagnosis

When the signs and symptoms make you suspect AHP, consider ordering all of the following random (spot) urine tests⁴



PBG¹¹
CPT Code: 84110*



ALA¹¹
CPT Code: 82135*



Porphyrins¹¹
CPT Code: 84120*

Substantial elevation of urinary PBG is a hallmark indicator of the 3 most common 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.

Testing for AHP: CPT Codes and Laboratory Results^{1,11,12}

Random (Spot) Urine [†] Test	CPT Code	Laboratory Results by AHP Type During an Attack			
		Acute Intermittent Porphyria	Hereditary Coproporphyria	Variegate Porphyria (VP)	ALAD-Deficiency Porphyria (ADP)
Porphobilinogen (PBG) [‡]	84110	Increased	Increased	Increased	No Increase
Delta-Aminolevulinic Acid (ALA) [‡]	82135	Increased	Increased	Increased	Increased
Porphyrins	84120	Increased uroporphyrin	Increased coproporphyrin (COPRO)	Increased COPRO	Increased COPRO

[†]Tests are more accurate when normalized per gram of creatinine and when sample is collected during acute episodes. 24-hour urine collection is generally not required. Additional testing (genetic or biochemical) may be required to determine AHP type (AIP, HCP, VP, or ADP).^{4,13}

[‡]ALA and PBG should be normalized to creatinine in order to adjust for differences in the degree of urinary concentration.¹³

Practical Considerations for Testing for AHP

- Lab tests for urine porphyrins do not include ALA/PBG¹⁴
- ALA levels are substantially elevated during symptomatic periods for all 4 AHP types, while PBG is substantially elevated for only the 3 most common forms (excluding ADP), but both ALA and PBG levels may decrease between attacks^{5,11,12,13}
- Tests are more accurate when normalized per gram of urine creatinine⁴
- Urine color can turn dark red or purple when exposed to light^{6,15}
- Samples should typically be light protected and frozen or refrigerated based on laboratory specifications. Short delays in refrigeration/freezing should not cause false-negative results^{4,16}

For the most up-to-date testing and protocol information, please refer to your local laboratories for specific requirements and guidance

CPT=Current Procedural Terminology.

⁵ALA and PBG are porphyrin precursors that occur naturally in the heme biosynthesis pathway in the liver but reach toxic levels in patients with symptomatic AHP.^{6,12}

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