

Use this discussion guide to help keep track of your signs and symptoms, and be sure to select all options that apply to your experience. During your next health visit, present this guide to your doctor to discuss if you should be tested for AHP.

1. Have you had severe, unexplained pain for more than one day in these areas?

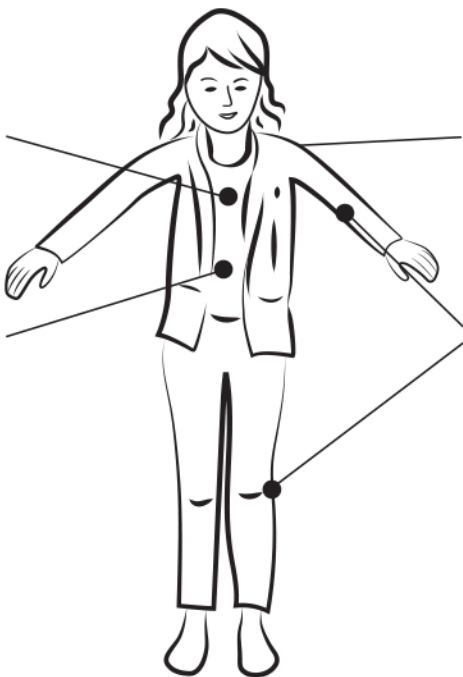
Circle where you have experienced this pain and describe any details using the lines below.¹

CHEST
(Describe)

BACK
(Describe)

BELLY
(Describe)

LIMBS
(Describe)



2. Have you experienced any of these signs and symptoms? Check all that apply¹⁻⁵:

Limb weakness or pain	Confusion	Abdominal pain	Dark or reddish urine
Numbness	Anxiety	Pain in back or chest	Low blood sodium
Fatigue	Seizures	Nausea and vomiting	
Tiredness	Insomnia	Lesions or blisters on sun-exposed skin*	
Paralysis	Hallucinations	Rapid heart rate	
Respiratory paralysis	Depression	High blood pressure	
Sensory loss	Constipation or diarrhea		

How long have you been experiencing these symptoms?

Have your symptoms ever required you to go to the hospital?
Yes No

*Hereditary coproporphyria and variegate porphyria.

Please write down any additional information you feel may be important to tell your doctor:

The acute hepatic porphyria (AHP) discussion guide

Start the conversation with your doctor

3. Have you had any of the following diagnoses or surgeries? Check all that apply:



Gastrointestinal disorders⁵⁻⁷

Irritable bowel syndrome (IBS)
Acute gastroenteritis with vomiting
Hepatitis



Neurological/neuropsychiatric disorders^{5,6}

Fibromyalgia
Guillain-Barré syndrome
Psychosis



Gynecological disorders⁶

Endometriosis



Abdominal conditions requiring surgery⁵

Appendicitis (inflammation of the appendix)
Cholecystitis (inflammation of the gallbladder)
Peritonitis (inflammation within the abdomen)
Intestinal occlusion (intestinal blockage)

After surgery, do you still have the same severe, unexplained pain?

Yes

No

Not applicable

4. Have symptoms started within recent days after exposure to any of the following?

Check all that apply¹:



SOME MEDICATIONS



HORMONE CHANGES

including levels of estrogen and progesterone. These hormones fluctuate the most during the 2 weeks before a woman's menstrual cycle begins.



DRINKING ALCOHOL



SMOKING



STRESS CAUSED BY:

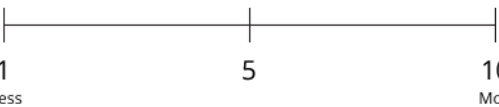
-Infections
-Surgery
-Physical exhaustion
-Emotional exhaustion



FASTING or extreme dieting

5. Have your symptoms disrupted parts of your life? Check all that apply^{8,9}:

Sleep Work Eating Socializing/Planning
Memory/Clear thinking Completing tasks
Maintaining energy Other: _____

How disruptive? 
How frequently? Daily Weekly Monthly Yearly

6. Has anyone in your family been previously diagnosed with a type of AHP?

Acute intermittent porphyria (AIP)	Variegate porphyria (VP)	Hereditary coproporphyria (HCP)
ALAD-deficiency porphyria (ADP)	No	Unsure

Please write down any additional information you feel may be important to tell your doctor:

AHP refers to a family of rare genetic diseases characterized by potentially life-threatening attacks and, for some people, chronic debilitating symptoms that negatively impact daily functioning and quality of life.^{1,8} The two most common techniques a doctor uses to help determine if a person has AHP are a **urine test** and a **genetic test**. A diagnosis of AHP is based on clinical judgement:^{3,11}

Urine Test



- A urine test for PBG (porphobilinogen), ALA (delta-aminolevulinic acid), and porphyrin levels can help inform a diagnosis of AHP*
- It is recommended to have a urine test during or shortly after an attack
- Porphyrin analyses may help identify the specific type of AHP, but are not used alone to diagnose AHP

Genetic Test



- A genetic test using a blood or saliva sample may help to confirm a diagnosis or determine the specific type of AHP
- It can help rule out AHP if there is not a genetic mutation
- A genetic test can be useful for family members of people with AHP who want to know if they carry the genetic mutation. Not everyone with a genetic mutation suggesting AHP will have symptoms⁵

*PBG and ALA are substances that are produced when the liver makes heme. Increased levels of PBG and ALA can become toxic and have been associated with the symptoms and attacks of AHP.^{2,10}

One genetic testing option:

Doctors can request no-charge genetic testing through the Alnylam Act[®] program for patients meeting certain criteria. While the program is sponsored by Alnylam Pharmaceuticals, all services are performed by independent third parties.

For more information and program rules, download a brochure at AlnylamActAHP.com.

AlnylamAct[®]

References: 1. Anderson KE, Bloomer JR, Bonkovsky HL, et al. *Ann Intern Med.* 2005;142(6):439-450. 2. Pischik E, Kauppinen R. *Appl Clin Genet.* 2015;8:201-214. 3. Balwani M, Wang B, Anderson KE, et al. *Hepatology.* 2017;66(4):1314-1322. 4. Harper P, Sardh E. *Expert Opin Orphan Drugs.* 2014;2(4):349-368. 5. Ventura P, Cappellini MD, Biolcati G, Guida CC, Rocchi E; Gruppo Italiano Porfiria (GriP). *Eur J Intern Med.* 2014;25(6):497-505. 6. Ko JJ, Murray S, Merkel M, et al. Poster presented at: American College of Gastroenterology Annual Scientific Meeting; October 5-10, 2018; Philadelphia, PA. 7. Alfadhel M, Saleh N, Alenazi H, Baffoe-Bonnie H. *Neuropsychiatr Dis Treat.* 2014;10:2135-2137. 8. Simon A, Pompilus F, Querbès W, et al. *Patient.* 2018;11(5):527-537. 9. Naik H, Stoecker M, Sanderson SC, et al. *Mol Genet Metab.* 2016;119(3):278-283. 10. Bissell DM, Anderson KE, Bonkovsky HL. *N Engl J Med.* 2017;377(21):2100-2101. 11. Bissell DM, Wang B. *J Clin Transl Hepatol.* 2015;3(1):17-26.