

# The Acute Hepatic Porphyria (AHP) Discussion Guide

## Start the conversation with your doctor

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.<sup>1</sup>

**CHEST**  
(Describe)

**BACK**  
(Describe)

**BELLY**  
(Describe)

**LIMBS**  
(Describe)

### 2. Have you experienced any of these signs and symptoms? Check all that apply:<sup>1,5</sup>

- |  |  |   |  |
|--|--|---|--|
| <input type="checkbox"/> Limb weakness or pain | <input type="checkbox"/> Confusion                 | <input type="checkbox"/> Unexplained Abdominal pain               | <input type="checkbox"/> Dark or reddish urine |
| <input type="checkbox"/> Numbness              | <input type="checkbox"/> Anxiety                   | <input type="checkbox"/> Pain in back or chest                    | <input type="checkbox"/> Low blood sodium      |
| <input type="checkbox"/> Fatigue               | <input type="checkbox"/> Seizures                  | <input type="checkbox"/> Nausea and vomiting                      |  |
| <input type="checkbox"/> Tiredness             | <input type="checkbox"/> Insomnia                  | <input type="checkbox"/> Lesions or blisters on sun-exposed skin* |  |
| <input type="checkbox"/> Paralysis             | <input type="checkbox"/> Hallucinations            | <input type="checkbox"/> Rapid heart rate                         |  |
| <input type="checkbox"/> Respiratory paralysis | <input type="checkbox"/> Depression                | <input type="checkbox"/> High blood pressure                      |  |
| <input type="checkbox"/> Sensory loss          | <input type="checkbox"/> Constipation or diarrhoea |   |  |

\*Hereditary coproporphyrria and variegate porphyria only.

How long have you been experiencing these symptoms?

\_\_\_\_\_

Have your symptoms ever required you to go to the hospital?

Yes     No

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

\_\_\_\_\_

\_\_\_\_\_

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



#### Gastrointestinal disorders<sup>5-11</sup>

- Irritable bowel syndrome (IBS)
- Acute gastroenteritis with vomiting
- Hepatitis
- Crohn's disease



#### Neurological/neuropsychiatric disorders<sup>5-11</sup>

- Fibromyalgia
- Guillain-Barré syndrome
- Psychosis



#### Gynecological disorders<sup>5-11</sup>

- Endometriosis



#### Abdominal conditions requiring surgery<sup>5</sup>

- 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 apply:<sup>1</sup>



- SOME MEDICATIONS**  
Talk to your healthcare professional to identify which drugs could be unsafe, possibly unsafe or safe for you.



- 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:<sup>12,13</sup>

- Sleep  Work  Eating  Socializing/Planning
- Memory/clear thinking  Completing tasks
- Maintaining energy  Other: \_\_\_\_\_

How disrupting?  1  5  10

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 coproporphyrin (HCP)
- ALAD-deficiency porphyria (ADP)  No  Unsure

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

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# How AHP is diagnosed

Acute hepatic porphyria (AHP) refers to a family of rare genetic diseases characterised by potentially life-threatening attacks and, for some people, chronic debilitating symptoms that negatively impact daily functioning and quality of life.<sup>1,12</sup> The two most common techniques a doctor uses to determine if a person has AHP are a **spot urine test** and a **genetic test**.<sup>3,15</sup>

## Spot Urine Test



- AHP can be diagnosed with a spot urine test of PBG (porphobilinogen), ALA (aminolevulinic acid), and porphyrin levels\*<sup>1,3,15</sup>
- A 24-hour urine collection is not recommended and may result in considerable delay in confirming the diagnosis<sup>18</sup>
- It is recommended to have a urine test during or shortly after an attack<sup>1,3</sup>
- Porphyrin analyses may help identify the specific type of AHP, but are not used alone to diagnose AHP<sup>3</sup>

## Genetic Test



- A genetic test using a blood or saliva sample may help to confirm a diagnosis or determine the specific type of AHP<sup>3</sup>
- It can rule out AHP if there is not a genetic mutation<sup>3</sup>
- AHP is a genetic inherited disease, so family members of someone who has AHP may also have inherited the altered gene responsible for the disorder. While most people with an altered gene may never have symptoms, they are however at risk of having an attack, or at risk of complications associated with elevated levels of ALA and PBG.<sup>16</sup> Knowledge of genetic risk of AHP may enable people to make informed decisions regarding lifestyle and medications with the intent to prevent attacks and complications of the disease.<sup>1</sup> Therefore, family members of someone who has AHP may want to talk with their doctor about genetic testing for AHP.<sup>17</sup>

\*PBG and ALA are substances that are produced when the liver makes haem. Increased levels of PBG and ALA can become toxic and have been associated with the symptoms and attacks of AHP.<sup>2,14</sup>

This information is intended for disease awareness purposes only. Nothing on the site constitutes individual medical advice. Individuals are advised to consult their physician or other appropriate HCP.

**Références :** **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.** Findley et al. *Contin Educ Anaesth Crit Care Pain.* 2012; 12(3):128-133. **9.** Sieg et al. *Gastroenterol.* 1991;29(11):602-605. **10.** Ali et al. *J Neurol Sci.* 2019;402:153-155. **11.** Mathavan et al. *J Hematol Thrombo Dis.* 2018;6(1):290. **12.** Simon A, Pompilus F, Querbes W, et al. *Patient.* 2018;11(5):527-537. **13.** Naik H, Stoecker M, Sanderson SC, et al. *Mol Genet Metab.* 2016;119(3):278-283. **14.** Bissell DM, Anderson KE, Bonkovsky HL. *N Engl J Med.* 2017;377 (21):2100-2101. **15.** Bissell DM, Wang BJ *Clin Transl Hepatol.* 2015;3(1):17-26. **16.** Ventura P, et al. *Intern Emerg Med.* 2009;4:297-308. **17.** Balwani M. *Clin Adv Hematol Oncol.* 2016;14:858-61. **18.** American Porphyria Foundation. Tests for porphyria diagnosis. <https://porphyriafoundation.org/for-patients/about-porphyria/testing-for-porphyria/tests-for-porphyria-diagnosis/>. Accessed 03 September 2020.