The Acute Hepatic Porphyria (AHP) Discussion Guide

Start the conversation with your doctor

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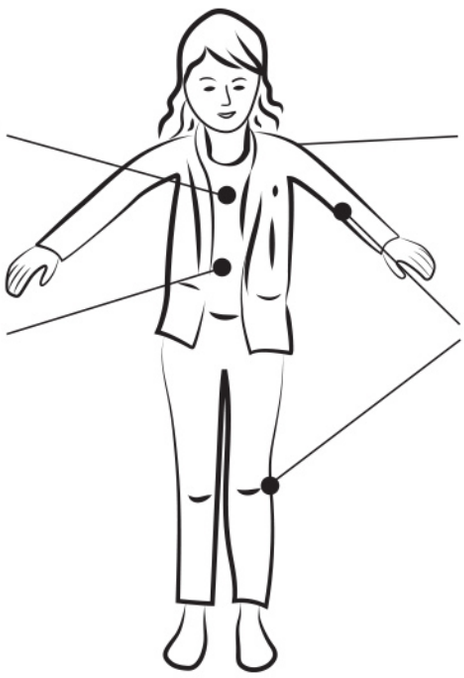
Acute Hepatic Porphyria

Use this discussion guide to keep track of your symptoms. Then present it to your doctor during your next visit to see if you should be tested for acute hepatic porphyria (AHP). Select all options that apply to your experience.

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]



2. Have you experienced any of these signs and symptoms? Check all that apply[1-5]:

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

*In hereditary coproporphyria and variegate porphyria types 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[5-7]

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

Neurological/neuropsychiatric disorders[5,6]

- ☐ Fibromyalgia
- ☐ Guillain-Barré syndrome
- ☐ Psychosis

Gynecological disorders[6]

- ☐ Endometriosis

Abdominal conditions requiring surgery[5]

- ☐ Appendicitis
- ☐ Cholecystitis
- ☐ Peritonitis
- ☐ Intestinal occlusion

After surgery, do you still have the same severe, unexplained pain? ☐ Yes ☐ No ☐ Not applicable

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

Check all that apply[1]:

☐ **TAKING CERTAIN MEDICATIONS**
(Please list medications)

☐ **HORMONE CHANGES**
including levels of estrogen and progesterone. These hormones fluctuate the most during the 2 weeks before a woman's menstruation begins.

☐ **DRINKING ALCOHOL**

☐ **SMOKING**

☐ **STRESS CAUSED BY:**
- –Infections
- –Surgery
- –Physical exhaustion
- –Emotional exhaustion

☐ **FASTING**
or low-carb diets

5. Have your symptoms disrupted parts of your life? Check all that apply[8,9]:

☐ Sleep ☐ Work ☐ School ☐ 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 coproporphyria (HCP)

☐ ALAD-deficiency porphyria (ADP) ☐ No ☐ Unsure

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

_____

_____

This discussion guide is not validated by any medical organization and does not replace the opinion of a trained medical physician.

Visit Porphyria.ca for more information about Acute Hepatic Porphyria.

How AHP is diagnosed

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Acute Hepatic Porphyria

Acute hepatic porphyria (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] People who may be experiencing symptoms and suspect AHP should ask their doctor to test for elevated PBG and ALA levels and porphyrins using simple spot urine tests. Below you can see how doctors arrive at an AHP diagnosis. After the urine tests, genetic tests can be used to confirm the specific type of AHP.[3]

1 Rule out other conditions

2 Suspect AHP signs and symptoms

3 Test urine PBG, ALA, and porphyrins[1,3,5,10]

(Biochemical test for PBG, ALA, and porphyrins)

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

It is recommended to have a urine test within 48 hours of symptom onset since PBG and ALA levels drop over time, increasing the chance of a false-negative result. Urine porphyrins is a nonspecific test and should not be used alone to diagnose AHP.[1,11]

*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.[2,10,12]

†Porphyrin analyses may help identify the specific AHP.[3]

4 Review the test results[3]

Positive—Your doctor may perform genetic testing or porphyrin analyses to confirm the type of AHP

Negative—If you and your doctor still suspect AHP, repeat the urine test during an attack, and/or consider genetic testing

No-charge genetic testing

Alnylam is sponsoring no-charge, third-party genetic testing for individuals who may carry a gene mutation known to be associated with AHP. The Alnylam Act® program was developed to reduce barriers to genetic testing to help people make more informed decisions about their health. While Alnylam provides financial support for this program, tests and services are performed by independent third parties. Healthcare professionals must confirm that patients meet certain criteria to use the program. Alnylam receives de-identified patient data from this program, but at no time does Alnylam receive patient identifiable information. Alnylam receives contact information for healthcare professionals who use this program. Genetic testing is available in the U.S. and Canada. Healthcare professionals who use this program have no obligation to recommend, purchase, order, prescribe, promote, administer, use or support any Alnylam product.

Your healthcare professional will need to sign up for the Alnylam Act® program in order for you to receive genetic screening at no charge. Find out more at AlnylamAct.com.

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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, Querbes 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. **12.** Gouya L, Bloomer JR, Balwani M, et al. Presented at: 2018 International Liver Congress; April 11-15, 2018; Paris, France.

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