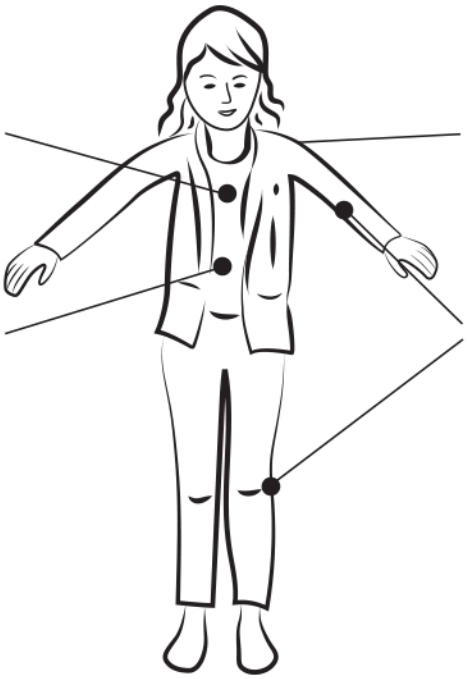
# The acute hepatic porphyria (AHP) discussion guide
Start the conversation with your doctor

Pinpoint® AHP
Acute Hepatic Porphyria

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 apply[1-5]:

- Limb weakness or pain
- Numbness
- Fatigue
- Tiredness
- Paralysis
- Respiratory paralysis
- Sensory loss
- Confusion
- Anxiety
- Seizures
- Insomnia
- Hallucinations
- Depression
- Constipation or diarrhea
- 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

*Hereditary coproporphyria and variegate porphyria.

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:

_____

_____

## 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

### Neurological/neuropsychiatric disorders[5,6]

- Fibromyalgia
- Guillain-Barré syndrome
- Psychosis

### Gynecological disorders[6]

- Endometriosis

### Abdominal conditions requiring surgery[5]

- 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[1]:

**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?

1 (Less) 5 10 (More)

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:

_____

_____

# How acute hepatic porphyria (AHP) can be diagnosed

Pinpoint® AHP
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] 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 symptoms[5]

*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, 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.



Visit PinpointAHP.com for more information about AHP.



Alnylam® PHARMACEUTICALS