The Acute Hepatic Porphyria (AHP) Discussion Guide

Start the conversation with your doctor

PORPHYRIA.ca
Porphyrie hépatique aiguë

If your doctor suspects AHP they should refer you to an AHP Specialist listed below

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] If someone in your family has been diagnosed with a form of AHP, please let your doctor know.

Use this guide to keep track of your symptoms, then present it to your family doctor at your next visit. If they suspect AHP based on your symptoms, they should refer you to an AHP Specialist listed below.

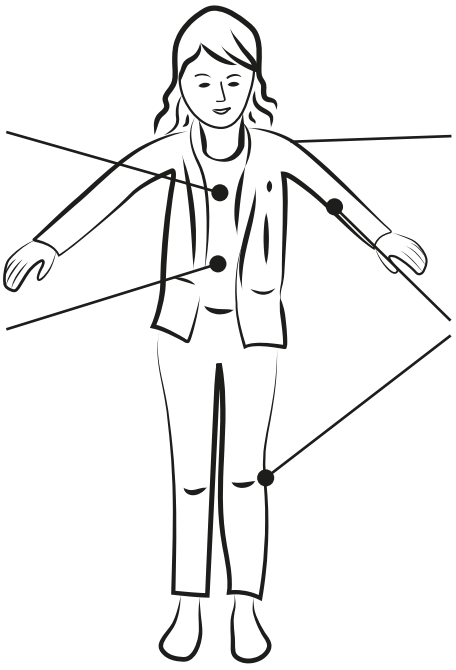
1. The below signs and symptoms can be associated with AHP. Please check all that apply:[1-15]

- ☐ Limb weakness or pain
- ☐ Numbness
- ☐ Fatigue
- ☐ Paralysis
- ☐ Respiratory paralysis
- ☐ Sensory loss
- ☐ Confusion
- ☐ Anxiety
- ☐ Seizures
- ☐ Insomnia
- ☐ Hallucinations
- ☐ Depression
- ☐ Constipation or diarrhea
- ☐ Unexplained abdominal pain
- ☐ Pain in back or chest
- ☐ Nausea and/or 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 types only.

2. Pain is a very common symptom of AHP, either general or specific.

Please circle areas on the diagram where you have experienced pain and describe the details of your pain below.



CHEST
(Describe)
_____
_____

BACK
(Describe)
_____
_____

BELLY
(Describe)
_____
_____

LIMBS
(Describe)
_____
_____

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.

Alnylam PHARMACEUTICALS

If your doctor suspects AHP they should refer you to an AHP Specialist listed below

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
- ☐ Choleocystitis
- ☐ Peritonitis
- ☐ Intestinal occlusion

If you had surgery, did or do you still have the same severe, unexplained pain? ☐ Yes ☐ No ☐ Not applicable

4. Often times symptoms can be triggered, worsened or more pronounced in the days following certain life events or activities.[1]

Below are some common triggers, check all that apply:

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

☐ **HORMONE CHANGES**
Including levels of estrogen or 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. People with AHP often need to seek medical attention from their family physician or an acute care facility (i.e., urgent care or hospital) multiple times, usually associated with an episode of more severe symptoms:[1,11-13]

Have you sought out help at a medical facility (family doctor, hospital, walk-in or acute care clinic) four or more times in a year for your symtoms? ☐ Yes ☐ No

On a scale of 1 to 10, how disruptive have your symptoms been for daily life?

1 5 10

How frequently do your symptoms disrupt your daily life? ☐ Daily ☐ Weekly ☐ Monthly ☐ Yearly

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.

If your doctor suspects AHP they should refer you to an AHP Specialist listed below

The symptoms outlined above could indicate Acute Hepatic Porphyria (AHP). If your patient has had unexplained severe abdominal pain – 92% of patients report abdominal pain[1,5,12-13] – plus one or more of the other symptoms referred to in this document, they may have AHP.[1] If a member of your patient's family has been diagnosed with any form of AHP and they have any of the above-mentioned symptoms, they could also have AHP. Below is a list of physicians that have significant experience diagnosing and treating this ultra-rare disease.

Quebec:

Dr. Jean Pierre Routy (MD, FRCPC)
Clinical Director of the Chronic Viral Illness Service
Division of Hematology and Chronic Viral Illness Service
Professor of Medicine, McGill University
Director, Réseau SIDA et maladies infectieuses FRQ-S
McGill University Health Centre: Glen site
1001 Blvd. Décarie, Montreal, QC H4A 3J1
Tel: 514-843-1558, Fax: 514-843-1418

Dr. Brian M. Gilfix (MDCM, PhD, FRCPC, DABCC, FACB)
Supervision of Testing for Acute and Chronic Porphyria
Consultant for Acute and Chronic Porphyria
Associate Professor, Division of Medical Biochemistry
MUHC-Royal Victoria Hospital
1001 Blvd. Décarie, Montreal, QC H4A 3J1
Tel: 514-934-1934 ext. 34410, Fax: 514-843-1499

Dr. Alan O'Brien (MD, FRCPC, FCCMG)
Clinical geneticist, CHUM
Assistant Clinical Professor, Université de Montréal
B05-043, 1000 rue St-Denis, Montreal, QC H2X 0C1
Tel: 514-890-8104

Ontario:

The Ottawa Hospital
General Campus Benign Hematology Program
501 Smyth Road, Ottawa, ON K1H 8L6
Fax: 613-737-8861

Dr. Michael Scott (MD, FRCPC, MHPE) & Dr. Michelle Sholzberg (MD, FRCPC, MSc)
Division of Hematology Oncology
Department of Medicine, St. Michael's Hospital
30 Bond St, Toronto, ON M5B 1W8
Tel: 416-864-5389, Fax: 416-864-3055

Dr. Margaret Elizabeth Turnbull (MD, MSc, FRCPC)
Hematologist, Medical Director of Transfusion Medicine
Royal Victoria Regional Health Centre
201 Georgian Drive, Barrie, ON L4M 6M2
Tel: 705-728-9802 ext. 23200, Fax: 705-797-3098

Alberta:

Dr. Eliza Phillips (MD, FRCPC, FCCMG)
Clinical Assistant Professor, Department of Medical Genetics
Alberta Children's Hospital
28 Oki Drive NW, Calgary, AB T3B 6A8
Tel: 403-955-7587, Fax: 403-476-8752

Dr. Andrei Fagarasanu (MD, PhD, FRCPC)
Associate Clinical Professor in Hematology at the University of Alberta
Vibe Medical Specialists
401, 11611-107 Ave NW, Edmonton, AB T5H 0P9
Tel: 780-496-1390, Fax: 780-496-1387

British Columbia:

Dr. Hayley Merkeley (MD, FRCPC)
Hematologist, Clinical Assistant Professor
St. Paul's Hospital/West Coast Hematology
411-1200 Burrard St, Vancouver, BC V6Z 2C7
Tel: 236-479-0498, Fax: 236-479-0498

Nova Scotia:

Dr. Ronald R. Yan (MD, FRCPC)
QEII Health Sciences Centre
1276 South Park St, Halifax, NS B3H 2Y9
Tel: 902-473-3447, Fax: 902-473-4600

Diagnosis: What Your Patient Should Expect

After ruling out other conditions, the specialist will test for elevated PBG (porphobilinogen) and ALA (aminolevulinic acid) levels, as well as porphyrins, using a simple spot urine test.[1-3,10-13,15] It is important that the urine test is conducted when you are experiencing these elevated symptoms in order to get an accurate measure of the severity.

Confirming Diagnosis

After the urine tests, a genetic test can be used to confirm the specific type of AHP.[1,2]
Genetic testing can also confirm a diagnosis if a false negative result from the urine tests is suspected. PBG and ALA levels drop over time, increasing the chance of a false negative result. For this reason, urine tests should ideally be done within 48 hours of symptom onset.[1,11]

No-Charge Genetic Testing

Genetic screening for AHP is available at no charge through the Alnylam Act program. Find out how you can access the free testing for your patients at **AlnylamAct.com**.

AlnylamAct®

References: 1. Anderson KE, et al. Ann Intern Med 2005;142:439-50. **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. Mo/ 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. **13**. Puy H, et al. Lancet 2010;375:924-37. **14.** Bonkovsky HL, et al. Ann Intern Med. 2005;142(6):439-450. **15.** Szlendak U, et al. Adv Clin Exp Med 2016;25:361-8

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

Alnylam® PHARMACEUTICALS