Family Health Tree

Acute Hepatic Porphyria (AHP) & Me

Map your family's history of Acute Hepatic Porphyria, one branch at a time, to determine who may be at risk.

AHP is inherited/genetic.

AHP refers to a family of rare genetic diseases, each being caused by a unique gene mutation, which inhibits production of a certain enzyme in the liver.[1] This results in the buildup of toxins called aminolevulinic acid (ALA) and porphobilinogen (PBG) in the liver, which are released throughout the body.[2] AHP attacks, chronic symptoms, and long-term complications occur when the nervous system reacts to the excess ALA and PBG.[2] Symptoms vary widely, but abdominal pain, nausea and vomiting, constipation, and confusion are common, usually appearing in individuals between 14 and 45 years old.[1, 3] AHP affects both men and women, although women are four times more likely to experience symptoms.[1]

AHP is generally passed down in an autosomal dominant pattern, which means a person only needs to inherit one copy of the affected gene from one parent in order to develop the disease risk.[1] When one parent carries an autosomal dominant mutation, any child will have a 50% chance of inheriting that mutation.[1]

Who is at risk? Are you at risk?

A family member may inherit the altered gene that causes AHP without ever developing symptoms.[1] Do you know anyone in your family, past or present, who has had symptoms that could be related to AHP but never a confirmed diagnosis? Knowledge of the 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.[3]

AHP: Acute Hepatic Porphyria; ALA: Aminolevulinic Acid; PBG: Porphobilinogen

This Family Health Tree assumes you have AHP. Working with family members to complete this tree could help them better understand AHP, its symptoms, how it is passed from one generation to the next, and identify who would benefit from genetic testing.

Let's grow your Tree. When you are ready, go to the Tree diagram on the next page.

Directions on Filling Out Your Family Health Tree

- Put yourself at the heart/center of your AHP Family Tree.
- List your symptoms using the number key (bottom left).
- At what age did your symptoms begin?
- At what age were you diagnosed with AHP?
- Next, add family members who are symptomatic or suspected carriers of AHP.

Need to expand your Tree? Simply add sheets of paper and draw in more boxes.

Completed your tree and wondering what to do next?

- if you feel ready, speak to your family
- if you need more support, speak to your healthcare provider / genetic counselor

Alicia, *AHP Patient and British Porphyria Association Volunteer*

1. Wang B et al. Hepatol Commun. 2018;3(2):193-206. 2. Gouya L, Ventura P, Balwani M, et al. Hepatology. 2020;71(5):1546-1558. 3. Anderson KE et al. Ann Intern Med. 2005 Mar 15;142(6):439-50.

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Map Your Family's History of Acute Hepatic Porphyria

Grandfather

Yes
- Experienced symptoms of AHP?
- ALA/PBG testing done?
- Genetic testing done?

Symptoms Number(s):

Age diagnosed | Age of onset of symptoms

Grandmother

Yes
- Experienced symptoms of AHP?
- ALA/PBG testing done?
- Genetic testing done?

Symptoms Number(s):

Age diagnosed | Age of onset of symptoms

Grandfather

Yes
- Experienced symptoms of AHP?
- ALA/PBG testing done?
- Genetic testing done?

Symptoms Number(s):

Age diagnosed | Age of onset of symptoms

Grandmother

Yes
- Experienced symptoms of AHP?
- ALA/PBG testing done?
- Genetic testing done?

Symptoms Number(s):

Age diagnosed | Age of onset of symptoms

*Aunt/Uncle

Yes
- Experienced symptoms of AHP?
- ALA/PBG testing done?
- Genetic testing done?

Symptoms Number(s):

Age diagnosed | Age of onset of symptoms

*Aunt/Uncle

Yes
- Experienced symptoms of AHP?
- ALA/PBG testing done?
- Genetic testing done?

Symptoms Number(s):

Age diagnosed | Age of onset of symptoms

Father

Yes
- Experienced symptoms of AHP?
- ALA/PBG testing done?
- Genetic testing done?

Symptoms Number(s):

Age diagnosed | Age of onset of symptoms

Mother

Yes
- Experienced symptoms of AHP?
- ALA/PBG testing done?
- Genetic testing done?

Symptoms Number(s):

Age diagnosed | Age of onset of symptoms

*Aunt/Uncle

Yes
- Experienced symptoms of AHP?
- ALA/PBG testing done?
- Genetic testing done?

Symptoms Number(s):

Age diagnosed | Age of onset of symptoms

*Aunt/Uncle

Yes
- Experienced symptoms of AHP?
- ALA/PBG testing done?
- Genetic testing done?

Symptoms Number(s):

Age diagnosed | Age of onset of symptoms

*Sibling/Cousin

Yes
- Experienced symptoms of AHP?
- ALA/PBG testing done?
- Genetic testing done?

Symptoms Number(s):

Age diagnosed | Age of onset of symptoms

*Sibling/Cousin

Yes
- Experienced symptoms of AHP?
- ALA/PBG testing done?
- Genetic testing done?

Symptoms Number(s):

Age diagnosed | Age of onset of symptoms

You

Yes
- Experienced symptoms of AHP?
- ALA/PBG testing done?
- Genetic testing done?

Symptoms Number(s):

Age diagnosed | Age of onset of symptoms

Partner/Spouse

Yes
- Experienced symptoms of AHP?
- ALA/PBG testing done?
- Genetic testing done?

Symptoms Number(s):

Age diagnosed | Age of onset of symptoms

*Sibling/Cousin

Yes
- Experienced symptoms of AHP?
- ALA/PBG testing done?
- Genetic testing done?

Symptoms Number(s):

Age diagnosed | Age of onset of symptoms

*Sibling/Cousin

Yes
- Experienced symptoms of AHP?
- ALA/PBG testing done?
- Genetic testing done?

Symptoms Number(s):

Age diagnosed | Age of onset of symptoms

Child

Yes
- Experienced symptoms of AHP?
- ALA/PBG testing done?
- Genetic testing done?

Symptoms Number(s):

Age diagnosed | Age of onset of symptoms

Child

Yes
- Experienced symptoms of AHP?
- ALA/PBG testing done?
- Genetic testing done?

Symptoms Number(s):

Age diagnosed | Age of onset of symptoms

Child

Yes
- Experienced symptoms of AHP?
- ALA/PBG testing done?
- Genetic testing done?

Symptoms Number(s):

Age diagnosed | Age of onset of symptoms

Symptoms:

1. Persistent and/or recurrent acute abdominal pain
2. Nausea and/or vomiting
3. Coloured/red urine
4. Constipation
5. Increased heart rate
6. Muscle weakness
7. Mood disorders/insomnia/confusion

Example

Claire

Yes
- ✓ Experienced symptoms of AHP?
- ALA/PBG testing done?
- ✓ Genetic testing done?

Symptoms Number(s): 2, 4, 6, 7

19 Age diagnosed | 18 Age of onset of symptoms

*Circle one and then draw a line to connect to relevant family members.

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Developed together with a steering committee consisting of European porphyria patient association leaders, acute hepatic porphyria (AHP) patients, caregivers, and health care professionals. Funded by Alnylam.

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