AlnylamAct®

Genetic Testing and Counseling Program*† for **Acute Hepatic Porphyria (AHP)** Offered at No Charge‡


Sean, living with AHP

Your doctor will help you determine if genetic testing through Alnylam Act® is the right choice for you.

*While program is sponsored by Alnylam Pharmaceuticals, all services are performed by independent third parties.
†Both genetic testing and genetic counseling are available in the US and Canada.
‡To patients, healthcare professionals, or payers.

About Acute Hepatic Porphyria (AHP)

What is 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. Attacks are commonly characterized by severe abdominal pain, vomiting, nausea, rapid heart rate, and constipation. During an attack, a person may also experience weakness, seizures, low sodium levels, and mental changes such as anxiety or confusion, or hallucinations in severe cases. There are four types of AHP that can be inherited differently:

There are 4 types of AHP that can be inherited differently

	Gene	Inheritance
AIP: Acute Intermittent Porphyria	***HMBS***	**Autosomal dominant**
VP: Variegate Porphyria	***PPOX***	**Autosomal dominant**
HCP: Hereditary Coproporphyria	***CPOX***	**Autosomal dominant**
ADP: ALAD-deficiency Porphyria	***ALAD***	**Autosomal recessive**

Depending on the type of AHP, the gene can be dominant or recessive

Autosomal Dominant: One pathogenic variant inherited from a single parent may be enough to cause disease in an individual

Autosomal Recessive: One pathogenic variant inherited from each parent (2 total) is needed to cause disease in an individual

Not everyone with a genetic mutation will have symptoms or attacks

What tests can help diagnose AHP?

People who may be experiencing symptoms and suspect AHP can ask their healthcare provider to test for AHP. The two most common techniques a doctor uses to help determine if a person has AHP are a urine test and a genetic test:

- **Urine Test:** Urine test for PBG (porphobilinogen), ALA (aminolevulinic acid), and porphyrin levels can help with the 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. Additional tests on plasma or stool samples may also be used to aid in diagnosis.
- **Genetic Test:** A genetic test using a patient specimen may help to confirm a diagnosis or determine the specific type of AHP. Genetic testing can tell a person if they carry a variant in a gene associated with AHP. Testing can be performed regardless of whether a person is currently experiencing attack symptoms. A genetic test can be useful for family members of people with AHP who want to know if they carry the genetic variant.

AlnylamAct®

How it works

1. **Get started**
   Ask your doctor about genetic testing for PH1 through Alnylam Act®
2. **Provide a sample for genetic testing**
   - Provide a sample using a PreventionGenetics sample collection kit (ask your doctor about sample collection options), **or**
   - Ask your doctor about the at-home specimen collection option offered by PreventionGenetics where you can collect a sample and send it back
3. **Pre-test genetic counseling** (optional)*
   Speak with a genetic counselor to learn more about genetic testing
4. **Review results**
   Results are ready in 3-4 weeks on average
   They will be sent directly to your doctor for review
5. **Post-test genetic counseling** (optional)*
   Speak with a genetic counselor to review your test results and what the results may mean for you and your family

*Both genetic testing and genetic counseling are available in the US and Canada.


Mary, living with AHP

Why is diagnosis important?

Early, accurate diagnosis of AHP may make a real difference in a person's ability to maintain their quality of life by avoiding the complications that can result from misdiagnoses and unnecessary surgeries. Testing as early as possible can help shorten the often lengthy time for AHP to get diagnosed. On average, it may take up to 15 years from symptom onset for a patient to receive a correct diagnosis of AHP.

Why is family screening important?

AHP is generally inherited through families, so a genetic test can help other family members find out if they may have AHP. Talking to a genetic counselor can help you and your family understand the risk of inheriting this disease.

Genetic testing process

If your doctor determines you are eligible, genetic testing is available through PreventionGenetics, a CLIA-certified independent genetic testing company.

1 Ask a healthcare professional to follow the instructions found at **www.preventiongenetics.com/sponsoredTesting/ahp**

2 Provide a DNA sample for genetic testing

3 Check back with your doctor within 3-4 weeks for your results

ASK YOUR DOCTOR
about providing a sample from your home for testing

Genetic counseling process

Alnylam has partnered with a third-party, Genome Medical, to provide no-charge genetic counseling services to any patient who enrolls in the Alnylam Act® program. Your doctor may order a pre-test and/or post-test genetic counseling session when they order your genetic test.

1 Genome Medical will contact you by phone or email to schedule your appointment

2 Speak with a genetic counselor to discuss the genetic testing process and/or your results*

3 Receive a summary of your genetic counseling session via email and access your summary in the Genome Medical portal **portal.genomemedical.com**

*If your test result is negative, Genome Medical will provide an educational video explaining the results.

GENETIC COUNSELORS are able to help you understand the testing process and your results. Genetic counselors can also assist with family variant testing.

AlnylamAct®

Make more informed decisions about your health

Talk with your doctor today about genetic testing for AHP

Why is genetic testing important for AHP?

Genetic testing can help identify your risk of AHP by looking at variants in genes associated with Acute Intermittent Porphyria (*HMBS*), Variegate Porphyria (*PPOX*), Hereditary Coproporphyria (*CPOX*), and ALAD-deficiency Porphyria (*ALAD*). Genetic testing may also help to shorten the time to diagnosis and prevent misdiagnoses.

What is genetic counseling?

Genetic counseling can help you and your family members learn more about AHP and the chances of inheriting the disease, and what may happen after a diagnosis.

How much does genetic testing and counseling cost through Alnylam Act®?

Alnylam Act® is a sponsored program, and the genetic testing and counseling services are offered to eligible patients at no charge.

Will any genetic or personal information be shared with Alnylam?

While Alnylam receives de-identified patient data from this program, at no time does Alnylam receive identifiable patient information. Third-party companies that support Alnylam Act® abide by applicable data privacy laws, including HIPAA and PIPEDA. Your information is safe and protected.

The Alnylam Act® program was created to provide access to genetic testing and counseling to patients as a way 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 may use healthcare professional contact information for research purposes
- Both genetic testing and genetic counseling are available in the US and Canada
- Healthcare professionals or patients who use this program have no obligation to recommend, purchase, order, prescribe, promote, administer, use, or support any Alnylam product
- No patients, healthcare professionals, or payers, including government payers, are billed for this program

Alnylam Canada ULC. 4 Robert Speck Parkway, Suite 1522 | Mississauga, Ontario L4Z 1S1
06, 2024
AHP-CAN-00136