



Genetic Testing and  
Counseling Program<sup>\*†</sup> for  
**Acute Hepatic Porphyria (AHP)**  
Offered at No Charge<sup>‡</sup>

Your doctor will help you  
determine if genetic testing  
through Anylam Act<sup>®</sup> is the  
right choice for you.



Sean, living  
with AHP

\*While program is sponsored by Anylam Pharmaceuticals, all services are performed by independent third parties.

†Genetic counseling only available in the U.S.

‡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         |
|--|-------------|---------------------|
| <b>AIP:</b> Acute Intermittent Porphyria | <i>HMBS</i> | Autosomal dominant  |
| <b>VP:</b> Variegate Porphyria           | <i>PPOX</i> | Autosomal dominant  |
| <b>HCP:</b> Hereditary Coproporphyria    | <i>CPOX</i> | Autosomal dominant  |
| <b>ADP:</b> ALAD-deficiency Porphyria    | <i>ALAD</i> | Autosomal recessive |

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

**Autosomal Dominant:** One parent with the mutation may be enough to cause the disease

**Autosomal Recessive:** Two parents with the mutation are needed to cause the disease

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 mutation 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 mutation.

# 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 mutations 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 through Alnylam Act<sup>®</sup> is available throughout the genetic testing process. Genetic counseling can help you and your family members learn more about AHP and the chances of inheriting the disease, how testing works, and what may happen after a diagnosis.

## How much does genetic testing and counseling cost through Alnylam Act<sup>®</sup>?

Alnylam Act<sup>®</sup> 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 provides financial support for this program, at no time does Alnylam receive identifiable patient information. The third-party companies that work with Alnylam abide by the Health Insurance Portability and Accountability Act (HIPAA), the same standards doctors use to protect your information.

## The Alnylam Act<sup>®</sup> 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 uses healthcare professional contact information for research and commercial purposes
- Genetic testing is available in the U.S. and certain other countries. Genetic counseling is available in the U.S.
- 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

## How it works

1

### Get started

Ask your doctor about genetic testing for AHP through Alnylam Act®

2

### Pre-test genetic counseling (optional)\*

Speak with a genetic counselor to learn more about genetic testing

3

### Provide a sample for genetic testing

- Provide a sample using an Invitae sample collection kit (ask your doctor about sample collection options), **or**
- Ask your doctor about the at-home specimen collection option offered by Invitae where you can collect a sample and send it back

4

### Review results

Receive results in 10-21 days. 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



### 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

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



- 1 Ask your doctor to follow the instructions found at [invitae.com/alnylam-act-ahp](https://www.invitae.com/alnylam-act-ahp)



- 2 Provide a blood, saliva, or buccal (cheek) swab sample for genetic testing



- 3 Check back with your doctor within 10-21 days for your results

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

Genetic testing through Alnylam Act<sup>®</sup> is provided through the Invitae Comprehensive Porphyrias Panel.

# Genetic counseling process

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If you are considering a genetic test for AHP, and you reside in the U.S., genetic counseling is available through InformedDNA, an independent genetic counseling company.



- 1 Call 1.888.475.3128 to schedule your appointment



- 2 Provide your doctor's name, address, phone, and fax number



- 3 Talk to a genetic counselor about AHP\*

**GENETIC COUNSELORS** are able to help you  
understand the testing process and your results

\*A genetic counseling report will be sent directly to your doctor within 1-2 weeks.

AnylamAct ®