AlnylamAct®

Genetic Testing and Counseling Program*† for **Primary Hyperoxaluria Type 1 (PH1)** Offered at No Charge‡


Benson, living with PH1

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 U.S. and Canada.
‡To patients, healthcare professionals, or payers.

About primary hyperoxaluria type 1 (PH1)


John, living with PH1

What is PH1?

PH1 is a rare, inherited disease that causes overproduction of oxalate, a substance that is normally only present in small amounts. Oxalate is a waste product that cannot be further broken down or used by the body, and is primarily eliminated by the kidneys. **PH1 is the most common and severe type of the primary hyperoxalurias.**

Why is diagnosis important?

Testing as early as possible can help shorten the often long time it takes for PH1 to get diagnosed. Adults can wait several years or more between their first symptom and receiving a PH1 diagnosis. This is important because the disease can progress in ways that aren't predictable and could lead to the development of kidney stones and chronic kidney disease.

What tests can help diagnose PH1?

While a healthcare professional will typically use a urine test to measure the urine oxalate excretion level, in a diagnostic workup of individuals suspected of primary hyperoxaluria, genetic testing can help confirm a diagnosis of PH1.

Why is family screening important?

PH1 is inherited through families. **If someone has PH1, their siblings have a 25% chance of also having the disease.**[1] It is important that siblings of a person with PH1 and other at-risk family members consider getting tested with a genetic test. Consultation with a geneticist or certified genetics counselor is recommended.

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How it works


Eloise, living with PH1

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

Pre-test genetic counseling (optional)*
Speak with a genetic counselor to learn more about genetic testing

3 **Review results**
Results are ready in 3-4 weeks on average. They will be sent directly to your doctor for review

4 **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.
**Genetic counseling after the testing is complete may be in the form of a one-on-one telephone visit or an educational video.

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

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 your doctor to follow the instructions found at **www.preventiongenetics.com/sponsoredTesting/ph1**

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 After the appointment, you can download a summary of your genetic counseling session to review and share with your healthcare provider

*Genetic counseling after the testing is complete may be in the form of a one-on-one telephone visit or an educational video.

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

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Make more informed decisions about your health

Why is genetic testing important for PH1?

Because PH1 is a genetic disease, genetic testing can help identify your risk and can help confirm a diagnosis of PH1 by looking for mutations in the *AGXT* gene. 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 PH1 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.

Talk with your doctor today about genetic testing for PH1

Reference: 1. Cochat P, Rumsby G. *N Engl J Med*. 2013;369(7):649-658.

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