



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 Alylam Act[®] is
the right choice for you.**

^{*}While program is sponsored by Alylam Pharmaceuticals, all services are performed by independent third parties.

[†]Genetic counseling only available in the U.S.

[‡]To patients, healthcare professionals, or payers.

About primary hyperoxaluria type 1 (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.

How it works



Eloise, living with PH1

- 1 Get started**
Ask your doctor about genetic testing for PH1 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

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 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

Genetic testing process

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-ph1](https://www.invitae.com/alnylam-act-ph1)



- 2 Provide a blood, saliva, or buccal 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 counseling process

If you are considering a genetic test for PH1, 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 PH1*

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.

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 through Alnylam Act® is available throughout the genetic testing process. Genetic counseling can help you and your family members learn more about PH1 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®?

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 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 U.S. Health Insurance Portability and Accountability Act (HIPAA), the same standards doctors use to protect your information.



Talk with your doctor
today about genetic
testing for PH1

AnylamAct 

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

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