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

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.
†Genetic counseling only available in the US.
‡To patients, physicians, and payers.
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 with PH1 can wait several years or more between their first symptom and receiving a PH1 diagnosis, which could lead to the development of kidney stones and chronic kidney disease. The disease can progress at an unpredictable rate, which is why early diagnosis is important. **About 50% of PH1 patients may be undiagnosed.**

How is it diagnosed?
While a urine test to measure oxalate excretion is used in the workup of individuals with PH1, it does not confirm a PH1 diagnosis. A genetic test is a simple way to confirm a PH1 diagnosis through a blood or saliva sample to determine if they have the disease.

Why is family screening important?
PH1 is inherited through families. 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.

Alnylam Pharmaceuticals is sponsoring no-charge, third-party genetic testing and counseling for people who may carry gene mutations known to be associated with PH1 through Alnylam Act®.
Get started
Ask your doctor about genetic testing for PH1 through Alnylam Act®.

Pretest genetic counseling (optional)
Speak with a genetic counselor to learn more about genetic testing.

Provide a sample for genetic testing
• Provide a blood or saliva sample at your doctor’s office, or
• Ask your doctor to send an Invitae specimen collection kit to you at your home and provide a saliva sample and send it back to Invitae

Review results
Results are ready in 10-21 days and sent directly to your doctor for review.

Post-test genetic counseling (optional)
Speak with a genetic counselor to review your test results and what they 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 US and Canada. Genetic counseling is only available in the US
• 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 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, an independent genetic testing company.

1. Ask your doctor to follow the instructions found at invitae.com/alnylam-act-ph1
2. Provide a blood or saliva sample for genetic testing
3. Check back with your doctor within 10-21 days for your results

ASK YOUR DOCTOR about how you can provide a saliva sample from your own home.

Genetic counseling process

If you are eligible for genetic testing and you reside in the US, 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 number, 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. Feel free to speak with a counselor as many times as you would like.

*A report will be sent directly to your doctor within 1-2 weeks.
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 before and after genetic testing. 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®?
While Alnylam covers the cost of the program, the genetic testing and counseling services are provided at no charge to patients, physicians, or payers.

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 HIPAA, the same standards doctors use to protect your information.