The Alnylam Act® program was developed to reduce barriers to genetic testing and counseling 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 receives contact information for healthcare professionals who use this program.

Genetic testing is available in the U.S. and Canada. Genetic counseling is only available in the U.S. Healthcare professionals and patients who use this program have no obligation to recommend, purchase, order, prescribe, promote, administer, use or support any Alnylam product.
There are three types of primary hyperoxaluria (PH): Type 1, Type 2, and Type 3. They differ in their severity and in their genetic cause. Primary hyperoxaluria Type 1 (PH1) is the most common form and accounts for about 80% of all cases. Patients with PH1 typically have kidney or bladder stones with symptoms such as severe lower abdominal pain, pain when urinating, and blood in the urine. PH1 is a life threatening disease that can also lead to kidney failure.

All types of primary hyperoxaluria are inherited in an autosomal recessive manner: a mutation is inherited from each parent. Children born to the same parents have a 25% risk to be affected with primary hyperoxaluria.

Because primary hyperoxaluria is a rare disease with unspecific symptoms, it may not be diagnosed for several years.

For more information about primary hyperoxaluria, visit the Oxalosis and Hyperoxaluria Foundation at [https://ohf.org](https://ohf.org).
What Is Genetic Counseling?
Genetic counseling is a service that provides information and support to people who have, or may be at risk for, genetic diseases.

What Is Genetic Testing?
Genetic testing can tell a person of any age whether they carry a gene change associated with a predisposition to, or diagnosis of, primary hyperoxaluria.

There are three genes associated with primary hyperoxaluria Types 1, 2 and 3: AGXT, GRHPR, and HOGA1 genes respectively. Genetic testing can be performed at any time, regardless of whether the person is currently experiencing pain from a kidney or bladder stone.

Genetic Testing Process
If your health care provider determines that you are eligible, genetic testing is available in the U.S. and Canada through Invitae, an independent genetic testing company.

- Your health care provider needs to follow the instructions found here: www.invitae.com/alnylam-act-ph1
- You will be asked to provide a blood or saliva sample for genetic testing
- Results are sent directly to your health care provider within 2-3 weeks

Genetic Counseling Process
Individuals who have a diagnosis of primary hyperoxaluria, have a known family history, or who are undergoing a clinical evaluation and potential genetic testing for primary hyperoxaluria, are eligible for genetic counseling through InformedDNA, an independent genetic counseling provider. This service is available in the United States only.

- Call InformedDNA at 1.888.475.3128 to schedule your appointment*
- Talk to a genetic counselor trained in primary hyperoxaluria over the phone
- Report is sent directly to your health care provider within 1-2 weeks

*Callers will need to provide their health care provider’s name, address, phone, and fax number.

Talk with your health care provider and/or a genetic counselor about the benefits, risks, limitations, and potential implications of testing for primary hyperoxaluria.
FOR PATIENTS

References

Alnylam is a biopharmaceutical company developing a new class of innovative medicines. We have a core focus on therapeutics toward genetically defined targets for the treatment of serious, life-threatening diseases.

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For assistance with genetic testing, call Invitae at 1.800.436.3037
For assistance with genetic counseling, call InformedDNA at 1.888.475.3128