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 who use this program have no obligation to recommend, purchase, order, prescribe, promote, administer, use or support any Alnylam product.
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.

Your health care provider needs to follow the instructions found here: www.invitae.com/alnylam-act-ph1

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.

We encourage you to speak with your health care provider and/or a genetic counselor about the benefits, risks, limitations, and potential implications of testing for primary hyperoxaluria.

What is Primary Hyperoxaluria Type 1? 1-5
Primary hyperoxaluria is an inherited disease caused by a gene change that causes calcium oxalate to build up faster than the kidneys can excrete it. This results in recurrent kidney or bladder stones, often occurring at a very young age. Other parts of the body, including the nervous system, heart, retina, and skin, can also be damaged if excess calcium oxalate accumulates there.

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. Although PH1 can occur at any age, most people with PH1 are less than 10 years old. They have kidney or bladder stones, and may have other symptoms including kidney failure, severe lower-abdominal pain, pain when urinating, and blood in the urine.

All types of primary hyperoxaluria are inherited in an autosomal recessive manner; a changed gene 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 an extremely rare disorder, it may not be diagnosed for several years.

For more information about primary hyperoxaluria, visit the Oxalosis and Hyperoxaluria Foundation at https://ohf.org.
FOR PATIENTS

References


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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