

# AnylamAct<sup>®</sup>

## Anylam Sponsored Third-Party Genetic Testing and Counseling Programs for **Primary Hyperoxaluria Type 1 (PH1)**



Lucy, living with primary hyperoxaluria type 1

The Anylam Act<sup>®</sup> program was developed to reduce barriers to genetic testing and counseling to help people make more informed decisions about their health. While Anylam 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. Anylam receives de-identified patient data from this program, but at no time does Anylam receive patient identifiable information. Anylam 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 Anylam product.

**No  
Charge**  
Genetic Testing  
and Counseling

## Why screen your patients for primary hyperoxaluria?<sup>1-5</sup>

The primary hyperoxalurias – Type 1, Type 2, and Type 3 – are metabolic disorders caused by mutations in the *AGXT*, *GRHPR*, and *HOGA1* genes respectively, resulting in the deposition of calcium oxalate crystals in the kidneys and urinary tract. Compromised kidney function exacerbates the disease as the excess oxalate can no longer be effectively excreted, resulting in subsequent accumulation and crystallization in bones, eyes, skin, and heart, leading to severe illness and the risk of premature death. The disorder is characterized by recurrent nephrolithiasis and/or ureterolithiasis, often occurring in childhood or adolescence. About 50 percent of patients will have kidney failure by age 15, and about 80 percent will have end stage renal disease by age 30. Current treatment options for advanced disease are very limited and include frequent renal dialysis or combined organ transplantation of liver and kidneys.

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

**No Charge**  
Genetic Testing and Counseling

### Genetic testing and counseling may help to:

- Identify risk of disease for patients and their family members
- Shorten the time to diagnosis and prevent misdiagnoses
- Determine whether patients are eligible to participate in clinical trials
- Provide information about support resources such as patient advocacy organizations

### Genetic testing for Primary Hyperoxaluria Type 1

Testing can be ordered as a single gene (*AGXT*). It can also be ordered as part of a three-gene panel that distinguishes PH1, PH2, and PH3; or as part of a larger panel that includes additional genes associated with hereditary conditions that have symptoms that may overlap with primary hyperoxaluria.



Claire, living with primary hyperoxaluria type 1

Patients of any age with a suspected diagnosis or a confirmed family history of primary hyperoxaluria Type 1 may be tested through the Alnylam Act<sup>®</sup> program.

### Testing options for your patients

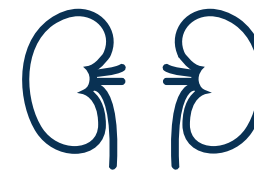
Invitae is the independent vendor providing this service.



#### Invitae Primary Hyperoxaluria (PH) Panel

Three-gene panel that includes next-generation sequencing with deletion/duplication testing, thereby detecting approximately 99% of disease-causing mutations in the following genes:<sup>1,2</sup>

PH Subtype	Gene
Primary Hyperoxaluria Type 1	<i>AGXT</i>
Primary Hyperoxaluria Type 2	<i>GRHPR</i>
Primary Hyperoxaluria Type 3	<i>HOGA1</i>



#### Invitae Nephrolithiasis Panel

Testing for 35 genes associated with inherited nephrolithiasis, including primary hyperoxaluria

### How to Order Genetic Testing Online\*



#### 1. Sign up online

Visit [www.invitae.com/alnylam-act-ph1](http://www.invitae.com/alnylam-act-ph1) to set up an account. Login to order a test. If needed, specimen (blood or saliva) collection kits can be ordered and typically take 2-3 days to arrive.



#### 2. Complete requisition and symptom checklist forms

In the Invitae Partner Code field, enter: **PH1**

Under Test Type, select: **Panel/Gene Test**

Under Test Selection, search for and select:

1. Invitae Primary Hyperoxaluria Panel
2. Invitae Nephrolithiasis Panel

Under billing information, select Institutional Billing and leave all fields blank to order testing at no charge



#### 3. Submit patient sample

Use a standard 4.0 mL lavender-top (EDTA) tube or saliva tube. Submit sample with completed forms. Specimen and shipping requirements are available on requisition form or online.



#### 4. Receive patient results

You will receive a notification email when results are ready, typically within 2-3 weeks.

\*For assistance with account set up, test ordering, or alternative ways to order testing and submit samples, call Invitae at 1.800.436.3037.

### How to Refer to Genetic Counseling

- InformedDNA is the independent vendor providing this service.
- Your patients can schedule a telephone-based genetic counseling session by calling 1.888.475.3128.
- Patients will need to reference the Alnylam Act<sup>®</sup> program and provide your contact information including your name, address, phone, and fax when scheduling their appointment.
- You will receive a detailed summary report of your patient's genetic counseling session.
- Genetic counseling is available before, during, or after genetic testing, and you do not need to set up an account for your patients to utilize this service.
- Genetic counseling is only available in the U.S.

## FOR PROVIDERS

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### References

1. Cochat P, Rumsby G. Primary hyperoxaluria. *N Engl J Med*. 2013;369(7):649-658.
2. Hopp K, Cogal AG, Bergstralh EJ, et al. Phenotype-Genotype Correlations and Estimated Carrier Frequencies of Primary Hyperoxaluria. *J Am Soc Nephrol*. 2015;26(10):2559-2570.
3. Hoppe B, Kemper MJ, Bökenkamp A, Portale AA, Cohn RA, Langman CB. Plasma calcium oxalate supersaturation in children with primary hyperoxaluria and end-stage renal failure. *Kidney Int*. 1999;56(1):268-274.
4. Zhao F, Bergstralh EJ, Mehta RA, et al. Predictors of Incident ESRD among Patients with Primary Hyperoxaluria Presenting Prior to Kidney Failure. *Clin J Am Soc Nephrol*. 2016;11(1):119-126.
5. Hoppe B, Bodo BB, Milliner D. "The Primary Hyperoxalurias." *Kidney Int*. 75.12 (2009): 1264–1271.



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 counseling, call InformedDNA at 1.888.475.3128