



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

**Consider genetic testing and
counseling for your patients;
Alnylam Act[®] provides one
option for eligible individuals**

^{*}While program is sponsored by Alnylam 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 progressive genetic disease that is potentially life-threatening and often presents with calcium oxalate kidney stones. Calcium oxalate crystals are insoluble and toxic and lead to serious disease manifestations, including recurrent kidney stones, nephrocalcinosis, progressive renal failure, and multiorgan damage from systemic oxalosis once the disease advances.¹⁻³

What is the role of genetic testing in PH1?

Due to the unpredictable progression of the disease, patients with PH1 benefit from early and accurate diagnosis. 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.^{2,4-6}



Eloise, living with PH1

Is family genetic testing important?

PH1 is an autosomal recessive disorder and may affect multiple individuals within a family. If you have a patient with PH1, consider genetic testing siblings and other at-risk family members. Consultation with a geneticist or certified genetics counselor is recommended to accelerate diagnosis.^{5,7}

Questions about variant of uncertain significance (VUS) results? Invitae's Clinical Consult Services are available to help aid in interpretation and resolution of VUS results. To discuss, please email clinconsult@invitae.com or call **1-800-436-3037**.

About Alnylam Act[®]

What is Alnylam Act[®]?

Alnylam Act[®] is a sponsored, no-charge, third-party genetic testing and counseling program for patients with a family history or suspected diagnosis of PH1. The Alnylam Act[®] program was developed to reduce barriers to genetic testing and counseling to help people make more informed decisions about their health.

What genes are tested through Alnylam Act[®]?

Genetic testing for PH1 offers 2 testing options: Invitae Primary Hyperoxaluria Panel (*AGXT*, *GRHPR*, and *HOGA1* genes) and Invitae Nephrolithiasis Panel (41 genes). The Invitae Nephrolithiasis panel tests for changes in the *AGXT* gene as well as for changes in other genes associated with nephrolithiasis that have symptoms that may overlap with PH1.

How much does genetic testing and counseling cost through Alnylam Act[®]?

Through the Alnylam Act[®] program, the genetic testing and counseling services are provided at no charge to patients, healthcare professionals, or payers, including government payers.

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

Getting Started

Once the decision to undergo genetic testing and counseling has been made:

Step 1 Determine eligibility for genetic testing*

Patients must meet one of the following criteria below:

- Family history of primary hyperoxaluria
 - OR
- Suspected diagnosis of primary hyperoxaluria with ONE OR MORE of the following symptoms:
 - Family history of primary hyperoxaluria OR
 - Adult (≥ 18 years old)- Elevated urinary oxalate OR Elevated plasma oxalate
 - Children (< 18 years old) with one of the following:
 - Failure to thrive AND impaired kidney function
 - Nephrolithiasis
 - Nephrocalcinosis
 - Elevated urinary oxalate OR Elevated plasma oxalate

Step 2 Order a genetic test



Start your order

- Request Invitae specimen collection kits to get started at [invitae.com/request-a-kit](https://www.invitae.com/request-a-kit)
- Complete the Invitae requisition form for Alnylam Act® (PH1), and fax it to **415-276-4164**, or insert the form in the specimen collection kit before shipment
- Note: You can also place an order through the online ordering portal at [invitae.com/alnylam-act-ph1](https://www.invitae.com/alnylam-act-ph1)



Submit patient sample

- Collect a sample using Invitae's specimen collection kit
- Follow the collection and shipping instructions inside the Invitae collection kit
- Note: Invitae offers the ability to send specimen collection kits directly to patients



Review results

- Receive results in 10-21 calendar days, on average
- Receive a notification email once the test results are ready. If you created an online account, you can view the status of your order by logging into your account

*Refer to the Alnylam Act® PH1 requisition form for full details of eligibility criteria.

Getting started (*continued*)

Step 3 Genetic Counseling (optional)



Refer patient for genetic counseling at any time*

- Instruct your patient to call InformedDNA at **1-888-475-3128** to schedule an appointment
- Patient may seek multiple genetic counseling sessions throughout the process, and counseling is available before, during, and after genetic testing



Prepare patient for the appointment

The patient will need to reference the Alnylam Act[®] program and provide your contact information (name, address, phone, and fax number) when scheduling the appointment



Receive results

A detailed summary report of your patient's genetic counseling session will be delivered to you via fax

*Genetic counseling only available in the U.S.



John, living with PH1

FOR QUESTIONS ABOUT GENETIC COUNSELING
Call InformedDNA at **1.888.475.3128**

FOR QUESTIONS ABOUT GENETIC TESTING
Contact Invitae at [invitae.com/contact](https://www.invitae.com/contact) or
1.800.436.3037

AnylamAct 

References: **1.** Hoppe B. *Nat Rev Nephrol.* 2012;8(8):467-475. **2.** Milliner DS, Harris PC, Cogal AG, Lieske JC. <https://www.ncbi.nlm.nih.gov/books/NBK1283/>. Updated November 30, 2017. Accessed October 16, 2019. **3.** Cochat P, Rumsby G. *N Engl J Med.* 2013;369(7):649-658. **4.** Ben-Shalom E, Frishberg Y. *Pediatr Nephrol.* 2015;30(10):1781-1791. **5.** Cochat P, Hulton SA, Acquaviva C, et al. *Nephrol Dial Transplant.* 2012;27(5):1729-1736. **6.** American Urological Association. <https://www.auanet.org/guidelines/kidney-stones-medical-mangement-guideline>. Published 2014. Accessed October 16, 2019. **7.** Edvardsson VO, Goldfarb DS, Lieske JC, et al. *Pediatr Nephrol.* 2013;28(10):1923-1942.

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