



# Genetic Testing and Counseling Program<sup>\*†</sup> for **Primary Hyperoxaluria Type 1 (PH1)** Offered at No Charge<sup>‡</sup>

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**Consider genetic testing and  
counseling for your patients;  
Alnylam Act<sup>®</sup> provides an option  
for eligible individuals**

<sup>\*</sup>While program is sponsored by Alnylam Pharmaceuticals, all services are performed by independent third parties.

<sup>†</sup>Genetic counseling only available in the US.

<sup>‡</sup>To patients, physicians, 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.<sup>1-3</sup>

## What is the role of genetic testing in PH1?

Due to the unpredictable progression of the disease, patients with PH1 benefit from an early and accurate diagnosis. While a urine test is used to measure the urine oxalate excretion in the workup of people with PH1, it does not confirm a PH1 diagnosis. A genetic test for mutations in the *AGXT* gene is a simple way to confirm a PH1 diagnosis.<sup>2,4-6</sup>

## 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.<sup>5,7</sup>



# Getting started

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

## Step 1 Determine eligibility for genetic testing\*

To be eligible for genetic testing through Alnylam Act<sup>®</sup>, patients must meet the requirements below:

- Family history of PH1, **or**
- Suspected diagnosis of PH1

## Step 2 Order a genetic test



### Start your order

- Request blood or saliva 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<sup>®</sup> (PH1), and fax it to **415-276-4164**, or insert the form in the Invitae 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 blood or saliva sample using Invitae's specimen collection kit
- Follow the collection and shipping instructions inside the Invitae kit
- Note: Invitae offers the ability to send saliva collection kits directly to patients for at-home saliva collection



### Review results

- Receive results in 10-21 calendar days, on average
- You will 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

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

\*Refer to the Alnylam Act<sup>®</sup> PH1 requisition form for full details about eligibility criteria.

# Getting started (*continued*)

## Step 3 Genetic counseling (optional)



### Refer your patient for genetic counseling at any time\*

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



### Prepare your patient for the appointment

The patient will need to reference the Alnylam Act<sup>®</sup> program and provide your contact information (name, address, phone number, 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 US.



Benson, living with PH1

**FOR ASSISTANCE WITH GENETIC COUNSELING,**  
call InformedDNA at **1-888-475-3128**.

**FOR ASSISTANCE WITH GENETIC TESTING,**  
contact Invitae at [invitae.com/contact](https://www.invitae.com/contact) or  
**1-800-436-3037**.

# About Alnylam Act<sup>®</sup>

## **What is Alnylam Act<sup>®</sup>?**

Alnylam Act<sup>®</sup> 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<sup>®</sup> program is designed 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<sup>®</sup>?**

Genetic testing for PH1 offers 2 testing options: Invitae Primary Hyperoxaluria Panel (*AGXT*, *GRHPR*, and *HOGA1* genes) and Invitae Nephrolithiasis Panel (35 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<sup>®</sup>?**

While Alnylam covers the cost of the program, the genetic testing and counseling services are provided at no charge to patients, physicians, or payers.

## **The Alnylam Act<sup>®</sup> 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

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**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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