Rose, living with an acute hepatic porphyria

Alnylam Act™

Alnylam Sponsored Third-Party Genetic Testing and Counseling Programs for the Acute Hepatic Porphyrias Offered at No Charge

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.

No Charge
Genetic Testing and Counseling

Alnylam Act™

Why Screen Your Patients for an Acute Hepatic Porphyria?[1-4]

The acute hepatic porphyrias—acute intermittent porphyria (AIP), variegate porphyria (VP), hereditary coproporphyria (HCP), and ALAD-deficient porphyria (ADP)—are characterized by acute, potentially life-threatening attacks and chronic debilitating symptoms that negatively impact patients' quality of life.

The acute hepatic porphyrias can resemble other conditions, and they are often misdiagnosed or remain undiagnosed for 15 years from symptom onset.

Common Misdiagnoses[1,5]

- Irritable Bowel syndrome
- Guillain-Barré syndrome
- Cholecystitis
- Appendicitis
- Hepatitis
- Endometriosis

Delays in diagnosis may lead to unnecessary surgeries and increased disease burden.

Biochemical Testing[3,6]

During a suspected porphyria attack, a urinary porphobilinogen (PBG) or aminolevulinic acid (ALA) test can enable the diagnosis of AIP, HCP, and VP. Urinary aminolevulinic acid (ALA) is the first-line test for ADP. These tests can be performed in a random urine sample and are more accurate when normalized per gram of urine creatinine—a 24-hour collection is not required. Specimen requirements are lab specific.

Biochemical testing is not available through the Alnylam Act™ program. For labs that perform biochemical testing, visit The Porphyrias Consortium website at: www.rarediseasesnetwork.org/cms/porphyrias

Genetic Testing for the Acute Hepatic Porphyrias (AHP)[7]

Genetic testing through the Alnylam Act™ program is available in a 4-gene panel that includes next generation sequencing with deletion/duplication testing, thereby detecting approximatively 99% of disease-causing mutations in the following genes:

AHP Subtype	GENE
Acute Intermittent Porphyria (AIP)	HMBS
Variegate Porphyria (VP)	PPOX
Hereditary Coproporphyria (HCP)	CPOX
ALAD-Deficient Porphyria (ADP)	ALAD

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 if patients are eligible to participate in clinical trials
- Provide information about support resources such as patient advocacy organizations

Who Is Eligible for Genetic Testing?

Patients must be at least 16 years old and meet eligibility from at least ONE of the criteria below:

Elevated (>ULN) urinary porphobilinogen (PBG) or aminolevulinic acid (ALA) levels

OR

Unexplained recurrent (more than one), prolonged (>24 hours) episodes of severe, diffuse (poorly localized) abdominal pain

AND: At least 2 of the following:

- Red to brownish urine, OR
- Known or suspected family history of an acute hepatic porphyria, OR
- Blistering skin lesions on sun-exposed areas, OR
- Peripheral nervous system manifestations occurring around the time of abdominal pain (i.e. motor neuropathy [paresis], sensory neuropathy [numbness, tingling, limb pain]), OR
- Central nervous system manifestations occurring around the time of abdominal pain (i.e. confusion, anxiety, seizures, hallucinations), OR
- Autonomic nervous system manifestations occurring around the time of abdominal pain (i.e. hyponatremia [Na <LLN], tachycardia, hypertension, nausea and vomiting, constipation)

How to Order Genetic Testing Online*

1. Sign up online

   Visit www.invitae.com/alnylam-act-ahp 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 eligibility criteria forms

   In the Invitae Partner Code field, enter: AHP

   Under Test Type, select: Panel/Gene Test

   Under Test Selection, search for and select: Acute Hepatic Porphyrias 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 setup, 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 eligible patients can schedule a telephone-based genetic counseling session by calling 1.888.475.3128
- Patients will need to reference the Alnylam Act™ 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.

References

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4. Anderson, et al. In: Proceedings from AASLD 2016;64(1):285A.
5. Kondo M, et al. Int J Hematol. 2004;79:448-456.
6. Bissell DM, et al. N Engl J Med. 2017;377(21):2101.
7. Whatley, et al. Clin Chem. 2009;55:1406–14.

Alnylam® PHARMACEUTICALS

Alnylam is a biopharmaceutical company developing a potential new class of innovative medicines. We have a core focus on therapeutics toward genetically defined targets for the treatment of serious, life-threatening diseases with limited treatment options for patients and their caregivers.
To learn more about Alnylam, please visit:
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