Alnylam Act™

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

Rose, living with an acute hepatic porphyria

Colin, living with an acute hepatic porphyria

No Charge
Genetic Testing

The Alnylam Act™ program was developed to reduce barriers to genetic testing 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. Healthcare professionals who use this program have no obligation to recommend, purchase, order, prescribe, promote, administer, use or support any Alnylam product.

What Is Porphyria?[1,2]

Porphyria is a group of disorders caused by abnormalities in the chemical steps that produce heme—a molecule in the body that is abundant in the blood, bone marrow, and liver. There are several types of porphyria that are characterized by the main site of the abnormality, such as the bone marrow (erythropoietic) or liver (hepatic), and by parts of the body affected, such as the skin (cutaneous) and/or nervous system (acute).

What Are the Acute Hepatic Porphyrias (Also Known as Acute Porphyrias)?[1-5]

There are four types of acute hepatic porphyria (AHP): acute intermittent porphyria (AIP), variegate porphyria (VP), hereditary coproporphyria (HCP), and ALAD-deficient porphyria (ADP).

The acute hepatic porphyrias are characterized by acute, potentially life-threatening attacks and chronic debilitating symptoms that negatively impact patients' quality of life.

Attacks are commonly characterized by severe abdominal pain, vomiting, nausea, rapid heart rate (tachycardia), and constipation. During an attack, a person may also experience muscle weakness or paralysis, seizures, low sodium levels (hyponatremia) and mental changes such as anxiety, confusion, or hallucinations in severe cases.

HCP and VP are classified as acute but can also have symptoms that affect the skin—specifically blistering skin lesions on sun-exposed areas. Skin symptoms can be present with or without attacks.

For more information about the acute hepatic porphyrias, visit the Canadian Association for Porphyria at http://canadianassociationforporphyria.ca.

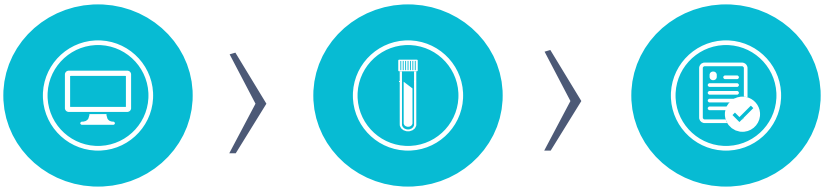
What Is Genetic Testing?[4,6,7]

Genetic testing can tell a person if they carry a mutation in a gene associated with a predisposition to, or diagnosis of, an AHP. Genetic testing can be performed regardless of whether a person is currently experiencing attack symptoms.

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.

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 order the Invitae Acute Hepatic Porphyrias panel and follow the instructions found here: www.invitae.com/alnylam-act-ahp

You will be asked to provide a blood or saliva sample for genetic testing of the four genes* associated with the AHPs

Results are sent directly to your health care provider within 2-3 weeks

We encourage you to speak with your doctor about the benefits, risks, limitations, and potential implications of testing for the acute hepatic porphyrias.

*HMBS for AIP, CPOX for HCP, PPOX for VP, and ALAD for ADP

References

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4. Anderson KE, et al. Ann Intern Med. 2005;142(6):439-50.
5. Anderson, et al. In: Proceedings from AASLD 2016;64(1):285A.
6. Bissell, et al. N Engl J Med. 2017;377:862-872.
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:
www.alnylam.ca

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For assistance with genetic testing, call Invitae at 1.800.436.3037