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.
What Are the Acute Hepatic Porphyrias (Also Known as Acute Porphyrias)?

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 American Porphyria Foundation at www.porphyriafoundation.org

What Is Porphyria?

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 Is Genetic Counseling?
Genetic counseling is a service that provides information and support to people who have, or may be at risk for, genetic diseases.

What Is Genetic Testing?
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.

Genetic Counseling Process
Individuals who have a diagnosis of an AHP, have a known family history, or who are undergoing a clinical evaluation and potential genetic testing for an AHP, are eligible for genetic counseling through InformedDNA, an independent genetic counseling provider. This service is available in the United States only.

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

Call InformedDNA at 1.888.475.3128 to schedule your appointment**

Talk to a genetic counselor trained in the acute hepatic porphyrias over the phone

Report is sent directly to your health care provider within 1-2 weeks

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

**Callers will need to provide their health care provider's name, address, phone, and fax number.
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.com

© 2018 Alnylam Pharmaceuticals, Inc.
300 Third Street | Cambridge, MA 02142 USA
05.2018

For assistance with genetic testing, call Invitae at 1.800.436.3037
For assistance with genetic counseling, call InformedDNA at 1.888.475.3128