

# An Overview of Genetic Counseling and Testing

Understand how your genes can increase your risk of developing hereditary ATTR (hATTR) amyloidosis



### Why genes are important

Genes carry information, called DNA, that determines traits and characteristics passed down to you from your parents. These genes control how you look, how your body works, and influence your skin, hair, and eye color. Sometimes there is a genetic variant, or change, in a gene that increases the likelihood of developing certain conditions like hATTR amyloidosis.

### hATTR amyloidosis is passed down through families

hATTR amyloidosis is a hereditary disorder that is caused by a genetic variant in the transthyretin (TTR) gene, and can affect several parts of the body, including the nerves, heart, and digestive system. If one parent has this disease, each child has a **50% chance of inheriting the genetic variant** that may cause this condition. A family member may inherit the TTR variant, but having the variant does not necessarily mean that they will develop hATTR amyloidosis.

Symptoms of hATTR amyloidosis are similar to those of other conditions, which can make it difficult for your doctor to diagnose. Therefore, genetic testing is an important part of the diagnostic process—as it determines whether you carry one of the **more than 120 gene variants** associated with the disease.



Genetic testing may help shorten the time to diagnosis and even prevent misdiagnosis.



## The 3 most common genetic variants that cause hATTR amyloidosis in the US

Although anyone can be at risk for this disease, it is more common for certain ethnicities, including people of **African, Brazilian, Irish, Japanese, Portuguese, and Swedish descent**. It's important to learn about your genetic history because it may help determine your risk for hATTR amyloidosis.

V1221

more common
in people of
African descent

**T60A** 

more common in people of **Irish** descent

**V30M** 

more common in people of Japanese and Portuguese descent

If your doctor thinks you may be at risk for hATTR amyloidosis, they may recommend you work with a genetic counselor.

### Who are genetic counselors?



As trained healthcare professionals, **genetic counselors** can help you learn more about hATTR amyloidosis and your personal risk of developing the condition. They can also help you understand the genetic testing process, and provide guidance and support.

#### Deciding to take action



Diagnosis of hATTR amyloidosis may involve various tests and procedures, but confirming a diagnosis may include genetic testing. Receiving a diagnosis can provide:

- A sense of relief from **receiving an accurate diagnosis**
- The ability to take action to make more informed decisions about your health
- The opportunity to better manage your symptoms earlier with your doctor
- Important knowledge that will help you inform your family about their own risk to aid them in their journey



## Genetic counseling and testing through Alnylam Act®

If your doctor determines you are eligible, Alnylam Act® offers third-party genetic counseling and testing for patients who may have hATTR amyloidosis—at no charge to patients, physicians, and payers, including government payers.

Genetic testing is available in the US and certain other countries. Genetic counseling is available in the US.

### What is the genetic counseling process<sup>a</sup>?



Call 1.888.475.3128 to schedule your appointment



Provide your doctor's
name, address, phone,
and fax number



Talk to a genetic counselor about hATTR amyloidosis<sup>b</sup>

<sup>a</sup>Genetic counseling is available through InformedDNA, an independent genetic counseling company.
<sup>b</sup>A detailed summary report of your genetic counseling session will be sent directly to your doctor within 1-2 weeks.

#### What is the genetic testing process<sup>c</sup>?



Ask your doctor
to follow the
instructions found at
www.invitae.com/
alnylam-act-ttr



Provide a sample, such as blood, cheek swab, or saliva for genetic testing.

Ask your doctor about how you can provide a cheek swab or saliva sample from your own home



Check back with your doctor within 10-21 days for your test results

You may speak to a genetic counselor multiple times throughout the process, either before, during, or after genetic testing.

<sup>&</sup>lt;sup>c</sup>Genetic testing is available through Invitae, an independent genetic testing company.



### Getting the right diagnosis is key

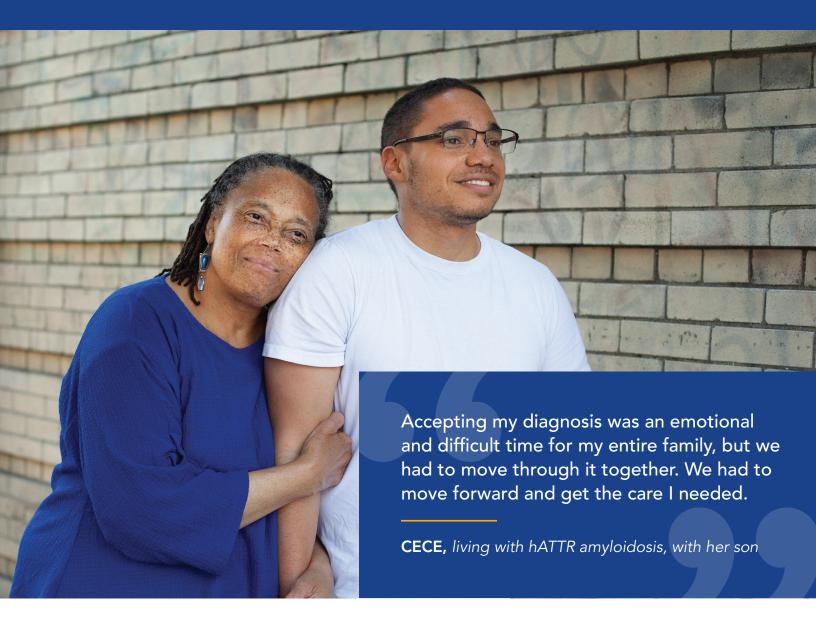
Alnylam Pharmaceuticals sponsors no-charge, third-party **genetic counseling and testing** for individuals who may carry one of the **120 or more gene variants** known to be associated with hATTR amyloidosis.

### Alnylam Act 🔄

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 US and certain other countries. Genetic counseling is 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 patients, healthcare professionals, or payers, including government payers, are billed for this program







Learn more about hATTR amyloidosis and genetic testing by visiting www.hATTRbridge.com.



