

# Hereditary ATTR (hATTR) amyloidosis

An introduction for you and your family to a rare condition





### How is hATTR amyloidosis passed down?

hATTR amyloidosis is an inherited condition—meaning **if one parent has hATTR amyloidosis**, **a child will have a 50% chance of inheriting a mutation that may cause this condition**.

Although anyone can be at risk for this condition, it is more common in certain ethnicities, such as people of African American, Portuguese, and Irish descent.

### What are the symptoms?

Symptoms of hATTR amyloidosis can vary among patients, even between family members, and can affect some parts of the body more than others.

Certain symptoms are typically seen early in the disease:

- Carpal tunnel syndrome
- Weakness
- Dizziness

- Numbness in hands or feet
- Diarrhea

Talk to your doctor if you think you may be experiencing hATTR amyloidosis symptoms or become aware of a family history.

### How hATTR amyloidosis (ama-loy-doh-sis) develops



Transthyretin (TTR), a naturally occurring protein made primarily in the liver, helps carry substances such as vitamin A.



In hATTR amyloidosis, a change (mutation) in the TTR gene can cause the protein to take on an abnormal shape (misfold).



Amyloid deposits

This change in shape causes the protein to build up in various parts of the body such as the nerves, heart, and digestive system. These abnormal proteins are called amyloid deposits.



Symptoms of hATTR amyloidosis

These amyloid deposits cause symptoms of the disease.



### Symptoms of hATTR amyloidosis

# Symptoms related to control over bodily functions

- Recurrent urinary tract infections (UTIs)
- Excessive sweating
- Dizziness upon standing
- Sexual dysfunction
- Nausea and vomiting
- Diarrhea
- Severe constipation
- Unintentional weight loss

#### Other symptoms

- Glaucoma
- Blurred or spotty vision
- Floaters in the eye
- Worsening dementia
- Stroke-like episodes
- Kidney dysfunction

# Symptoms related to the heart

- Fatigue
- Dizziness
- Shortness of breath
- Leg swelling (edema)

# Symptoms related to nerves in hands, feet, arms, and legs

- Tingling
- Numbness
- Carpal tunnel syndrome
- Burning pain
- Loss of sensitivity to temperature
- Loss of movement control
- Weakness

This is not a complete list of symptoms that may be experienced in patients with hATTR amyloidosis. Each patient has a different experience and you may not experience all of these symptoms, or you may not experience them at the same time. Symptoms of hATTR amyloidosis may worsen over time.

## How is hATTR amyloidosis diagnosed?

If your doctor believes you are at risk for hATTR amyloidosis, he or she may recommend:

• Cardiac and/or neurological tests • Genetic testing



Your doctor may also recommend you work with a genetic counselor, who can help you and your family learn more about the testing process and if a genetic test may be right for you. Genetic testing can help determine whether a person carries a genetic mutation associated with hATTR amyloidosis.

### Are there genetic testing options available? Alnylam Act<sup>®</sup>

Alnylam Pharmaceuticals is sponsoring no-charge, third-party genetic counseling and testing for individuals who may carry one of the 120 or more gene mutations known to be associated with hATTR amyloidosis.<sup>a</sup>

#### **Consumer genetic services**

Alnylam has teamed up with 23andMe to offer a Hereditary Amyloidosis (TTR-related) Genetic Health Risk Report<sup>b</sup> that can detect whether you have 1 of 3 genetic mutations known to cause hATTR amyloidosis. Our collaboration includes the Alnylam +myFamily program, which provides Health + Ancestry Service Kits at no charge to family members of 23andMe customers who are found to be carriers of certain genetic mutations associated with hATTR amyloidosis.

<sup>a</sup>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 and patients who use this program have no obligation to recommend, purchase, order, prescribe, promote, administer, use or support any Alnylam product.

<sup>b</sup>23andMe services provide information on a person's risk for certain diseases, but cannot provide a diagnosis. The Hereditary Amyloidosis (TTR-related) Genetic Health Risk Report provides information on the 3 most common TTR mutations in the United States. There are over 120 mutations associated with hATTR amyloidosis. If you have symptoms of hATTR amyloidosis or other symptoms you are concerned about, regardless of results reported by a 23andMe Health Risk Report, it is important to talk to your doctor as soon as possible. Only a doctor can confirm a diagnosis of hATTR amyloidosis.



There came a point when our entire family said 'enough.' We decided to arm ourselves with knowledge of hATTR amyloidosis. My family was fortunate that we found where to get help and support.

**SUE,** member of a family living with hATTR amyloidosis



Visit www.hATTRbridge.com for more information about hATTR amyloidosis.



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