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, and each child of one parent with hATTR amyloidosis has a 50% chance of inheriting the genetic variant, or change, that causes this condition. This change in the transthyretin (TTR) gene may also be referred to as a mutation.

Although anyone can be at risk for this disease, it is more common in certain ethnicities, such as people of African, Brazilian, French, Irish, Japanese, Portuguese, and Swedish descent.

What are the symptoms?

Symptoms of hATTR amyloidosis can vary widely among people with the condition and even within families. These symptoms can affect multiple parts of the body.

Certain symptoms typically appear early in the disease:

- Carpal tunnel syndrome
- Numbness in hands or feet
- Weakness
- Diarrhea
- Dizziness
- Unintentional weight loss

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

TTR is a naturally occurring protein made primarily in the liver and carries substances such as vitamin A.

In hATTR amyloidosis, a variant, or change, in the TTR gene causes the protein to take on an abnormal shape and misfold.

This change in shape causes the protein to build up in various parts of the body, including the nerves, heart, and digestive system. The collection of abnormal proteins is called amyloid deposits.

These amyloid deposits cause symptoms of the disease.
Symptoms of hATTR amyloidosis

Symptoms related to control over bodily functions
- Recurrent urinary tract infections (UTIs)
- Abnormal 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 thinks you may be at risk for hATTR amyloidosis, they 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 variant in the TTR gene associated with hATTR amyloidosis.

Are there genetic testing options available?

Alnylam Act®

Alnylam Pharmaceuticals is sponsoring 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.¹

Consumer genetic services

Alnylam has partnered with 23andMe to offer consumer genetic services that look for the 3 most common genetic variants associated with hATTR amyloidosis. This includes the +myFamily Program, which provides 23andMe Health + Ancestry Service Kits at no charge to eligible family members of 23andMe customers who are found to be carriers of genetic variants known to cause hATTR amyloidosis.²

¹While 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 certain other countries. Genetic counseling is 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. No patients, healthcare professionals, or payers, including government payers, are billed for this program.

²23andMe 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 variants in the United States. There are over 120 variants 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, living with hATTR amyloidosis

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