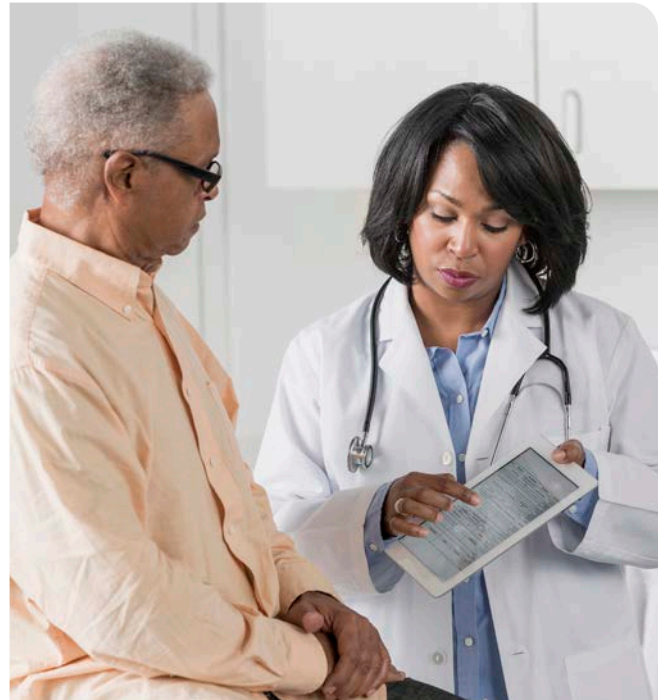


# What is hATTR amyloidosis?

Hereditary ATTR (hATTR) amyloidosis is an inherited condition. People who have it are born with a genetic variant affecting a protein called transthyretin (TTR), which is primarily made by the liver. Normally TTR helps transport vitamin A and thyroxine, a thyroid hormone, through the blood.

In hATTR amyloidosis, a variant in the TTR gene causes the TTR protein to take on an abnormal shape and misfold. The misfolded TTR can build up as amyloid in the heart, nerves, and other organs and tissues causing a variety of symptoms. hATTR amyloidosis is progressive and is associated with a diminished quality of life and life expectancy between 2.5 and 5 years after diagnosis.



## How prevalent is hATTR amyloidosis?

It's estimated that about 50,000 people worldwide have hATTR amyloidosis, although many researchers believe the actual number may be far higher. It can occur in all races and ethnicities, but it's more common in people of West African, Irish, or Portuguese ancestry. It has also been diagnosed in greater than expected numbers in Sweden and Japan.

## How does it affect the body?

The buildup of amyloid fibrils can begin years before symptoms begin. However, as hATTR amyloidosis progresses it can impact many organ systems and body parts. Although everyone experiences the progression of the disease differently, some signs and symptoms are more common than others, with some occurring earlier in the disease. Most people with hATTR amyloidosis will develop a combination of, but not every sign and/or symptom.

### Movement and Sensation Symptoms

- Reduced ability to perform activities of daily living
- Dizziness or fainting when standing up from a sitting or lying position (low blood pressure called orthostatic hypotension)

### Heart Symptoms

- Heart failure (including fatigue, shortness of breath, swelling in the legs, ankles and feet, reduced ability to exercise)
- Other heart problems (irregular heartbeats and rhythm disorders)

### Stomach and Digestive System Symptoms

- Nausea and vomiting
- Constipation, daily diarrhea or alternating bouts of constipation and diarrhea
- A feeling of fullness after eating a small amount of food (early satiety)
- Unexpected weight loss

### Eyes

- Floaters
- Glaucoma
- Vessel and papillary irregularities
- Dry eye syndrome

### Kidneys

- Excess protein in the urine
- Kidney failure

### Symptoms in Other Body Parts

- Bilateral carpal tunnel syndrome
- Swelling in the legs
- Narrowing of the spinal column (lumbar spinal stenosis)
- Sudden bicep tendon rupture
- Erectile dysfunction
- Excessive sweating
- Bladder problems

### Head and Central Nervous System Symptoms (less common)

- Headache
- Seizures, spasms, and/or lack of muscle control
- Dementia
- Stroke-like episodes

(continued)



### How do I know if I should get tested?

Talk to your health care professional about whether you should get tested for hATTR amyloidosis if you have a combination of the signs and/or symptoms above or any of the following:

- A family history of hATTR amyloidosis
- Peripheral neuropathy
- Heart failure with preserved ejection fraction (HFpEF) and increased left ventricular wall thickness
- Bilateral carpal tunnel syndrome
- Spinal stenosis
- Sudden biceps tendon rupture
- Previous orthopedic procedures
- Intolerance to some heart failure medications

### How is hATTR amyloidosis diagnosed?

Because hATTR amyloidosis affects many organs and body parts as well as causes a wide variety of symptoms, it can be challenging to diagnose.

However, there are several tests that can help determine if you have hATTR amyloidosis. Some of these common diagnostic tests include:

- Genetic testing
- Biopsy
- Blood test
- Urine test
- Bone scan
- Cardiac imaging
- Nerve conduction
- Eye exam

If your health care professional suspects you have hATTR amyloidosis, a biopsy may be ordered to confirm the presence of

amyloid deposits and identify the exact protein causing them. In conjunction with blood and urine testing, a type of bone scan called scintigraphy also can detect amyloid deposition in the heart, sometimes eliminating the need for a biopsy.

Blood tests can also provide information about how the amyloid fibrils are affecting your heart, kidney and liver.

Additional testing, including cardiac imaging like echocardiography and MRI, and nerve conduction tests, can determine how hATTR amyloidosis has impacted the heart and nervous systems.

If signs of heart, liver, kidney or other problems are discovered, your health care professional will likely start treating you for those conditions.

If initial diagnostic testing suggests you may have hATTR amyloidosis or if you have a family history of hATTR amyloidosis, your health care professional may refer you to a genetic counselor and/or order genetic testing to determine if you have the variant that causes the hereditary condition. If a parent has the variant, you have a 50% chance of having it, too. However, having a variant does not necessarily mean that you will develop hATTR amyloidosis.

### What are the next steps?

It's important to diagnose hATTR amyloidosis early so your health care team can initiate proper care and treatment.

**Talk with a trusted health care professional about the most appropriate treatment options for you.**

## HOW CAN I LEARN MORE?

- 1 Call 1-800-AHA-USA1 (1-800-242-8721), or visit [heart.org](http://heart.org) to learn more about heart disease and stroke.
- 2 Sign up for our monthly *Heart Insight* e-news for heart patients and their families at [HeartInsight.org](http://HeartInsight.org).
- 3 Connect with others sharing similar journeys with heart disease and stroke by joining our Support Network at [heart.org/SupportNetwork](http://heart.org/SupportNetwork).

### Do you have questions for your doctor or nurse?

Take a few minutes to write down your questions for the next time you see your health care professional.

For example:

**How is a genetic test conducted?**

**Should my family members get tested, too?**

## MY QUESTIONS:

We have many other fact sheets to help you make healthier choices to reduce your risk for heart disease, manage your condition or care for a loved one. Visit [heart.org/AnswersByHeart](http://heart.org/AnswersByHeart) to learn more.