

# Hereditary ATTR amyloidosis

A closer look at an inherited condition



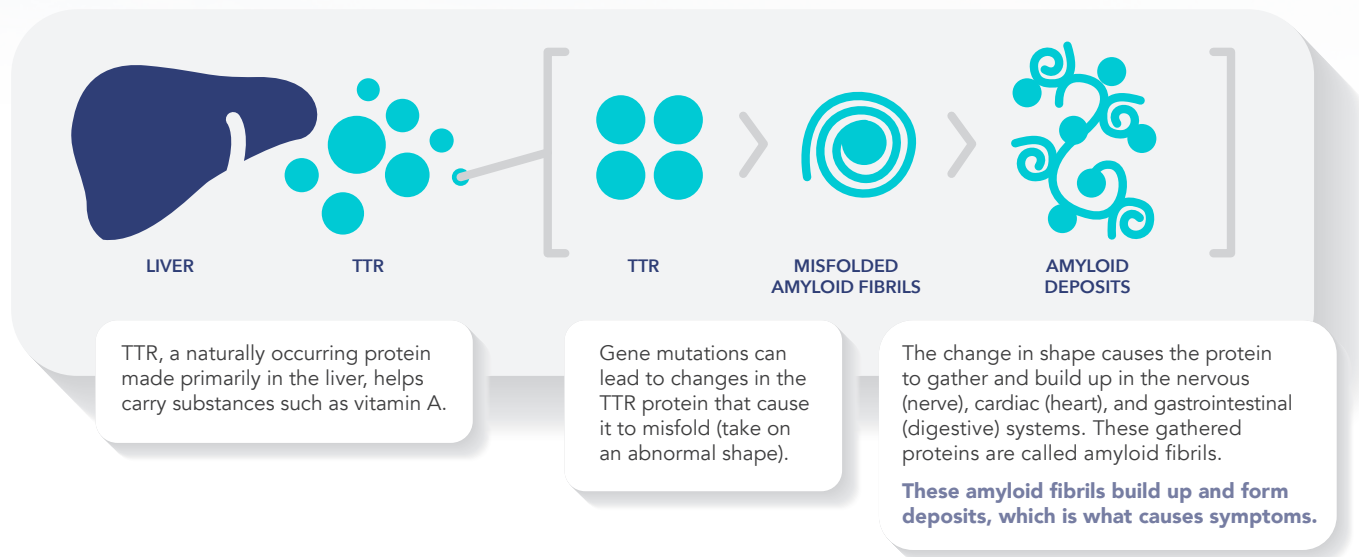
the   
**BRIDGE**  
hATTR amyloidosis

## What is hereditary ATTR (hATTR) amyloidosis?

hATTR amyloidosis is caused by a gene change (mutation) that affects the function of a protein in the blood called transthyretin (TTR). This protein is made primarily in the liver. hATTR amyloidosis is a rare condition that affects an estimated 50,000 patients worldwide.

Individuals with hereditary ATTR amyloidosis may have a range of symptoms that may seem unrelated. Symptoms can affect several parts of the body, including the nervous (nerve), cardiac (heart), and gastrointestinal (digestive) systems.

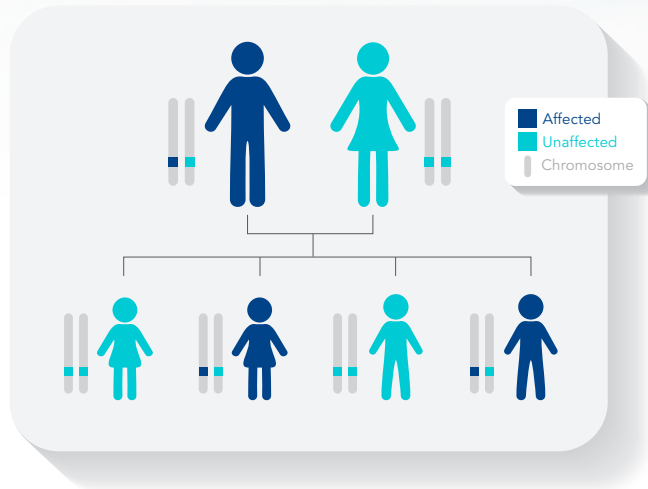
### How hATTR amyloidosis develops



## hATTR amyloidosis—an inherited condition

hATTR amyloidosis is passed down through affected family members, and is inherited in an autosomal dominant fashion, meaning a person only needs to inherit one copy of the affected gene from one parent in order to develop the condition.

Genes are located on structures known as chromosomes. Every individual has two copies of the TTR gene, one inherited from each parent. When one parent carries an autosomal dominant mutation, **any child will have a 50% chance** of inheriting that mutation.



A family member may inherit the TTR gene with a mutation, but having the mutation does not mean he or she will develop hATTR amyloidosis.



hATTR amyloidosis can be passed down to children, an important consideration for people who may be thinking about starting a family.

## hATTR amyloidosis can cause a range of symptoms

The symptoms of hATTR amyloidosis can vary widely among people with the same mutation and even within families, though some do see a pattern of symptoms develop. Different symptoms may appear at different times for each individual. The age that initial symptoms appear may vary, ranging from the mid-20s to the mid-60s.

### hATTR amyloidosis affects several parts of the body, including:



The peripheral nervous system, which is made up of nerves that branch out from the brain and spinal cord and communicate with the rest of the body including your arms and legs

- Polyneuropathy is caused by damage to the nerves of the peripheral nervous system, resulting in improper function



The cardiac system, which includes the heart and blood vessels, transports blood through veins and delivers oxygen to cells in the body

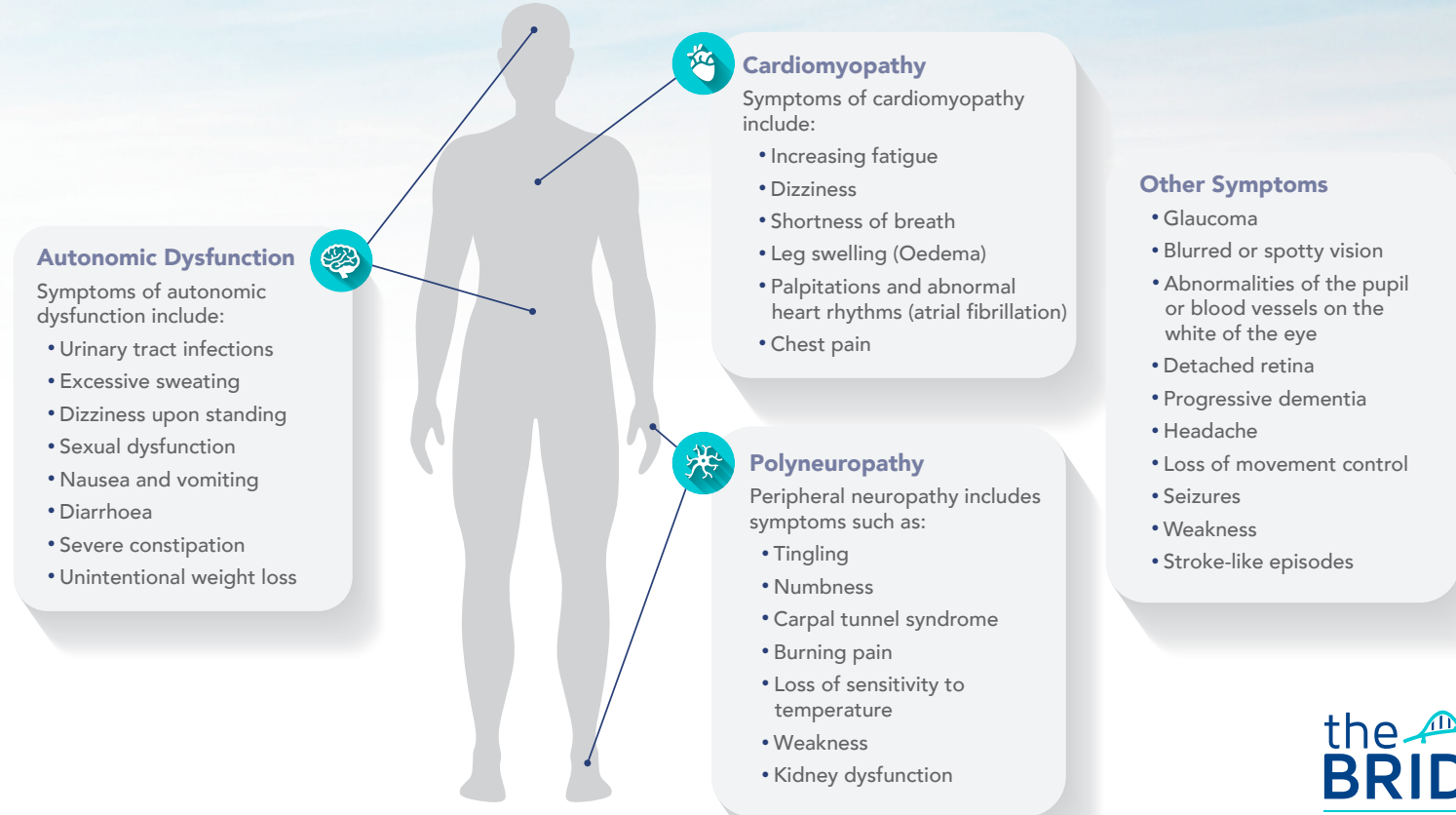
- Cardiomyopathy is a disease of the heart muscle that leads to heart failure



The autonomic nervous system, which is made up of nerves that connect the brain and spinal cord to organs such as the heart, stomach, and intestines, and helps to control bodily functions such as breathing, digestion, and heart rate

- Autonomic dysfunction occurs when the autonomic nervous system is not working correctly and may affect involuntary bodily functions

# The varying symptoms of hATTR amyloidosis



## Getting the right diagnosis is key

Misdiagnosis is common with hATTR amyloidosis because the symptoms can resemble those of other conditions. Learning about the symptoms of hATTR amyloidosis can help you identify them if they occur.

### Take the next step

If you experience symptoms or become aware of a family history, speak to your healthcare professional to determine the right plan of action. Because the condition is rare and affects different parts of the body, you may be referred to a specialist who is more familiar with hATTR amyloidosis. Even if you haven't experienced any symptoms but are aware of a family history, your healthcare professional may refer you to a genetic counsellor.

## Get the facts about hereditary ATTR amyloidosis

Learn about hATTR amyloidosis at [www.hATTRbridge.ca](http://www.hATTRbridge.ca)



“Right away, I wanted to know if I can do something about this.”

— Sandie,  
*living with hATTR amyloidosis*





## Notes

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