What is hATTR Amyloidosis?

Hereditary ATTR amyloidosis (hATTR) is an inherited condition (passed down through families) that often affects the liver, nerves, heart and kidneys. hATTR amyloidosis is characterized by the deposit of an abnormal protein called amyloid in multiple organs of the body where it should not be, which causes disruption of organ tissue structure and function. In hereditary amyloidosis, amyloid deposits most often occur in tissues of the nervous system, heart, and digestive tract.¹

hATTR amyloidosis is passed down through family members (genetically inherited). It is an autosomal dominant condition, meaning an individual needs to inherit only one copy of the mutated gene from one parent to develop the disease.² If one parent has hATTR amyloidosis, there is a 50% chance their child will inherit the mutation.

TTR amyloid fibrils form when the structure of a normally dissolvable TTR protein misfolds.¹

The misfolded proteins are sticky and clump together in tissues and between the body’s cells to form amyloid deposits.³

Amyloid deposits cause illness by damaging the structure and the function of the organs where they are found. They can affect almost any part of the body including the nerves, heart and GI tract.³

About 50,000 people worldwide have hATTR amyloidosis⁴

Patients often have to see 5+ doctors across different specialties before getting the right diagnosis

Often takes more than 4 years from symptom onset to a diagnosis
The first symptoms of hATTR amyloidosis typically appear between the mid-20s and the mid-60s, involving multiple tissues and organs and often seem unrelated. Because symptoms may be confused with more common conditions, hATTR amyloidosis can be hard to diagnose.¹

Patients often present with a cluster of one, two, three or more red-flag symptoms, including:

- **Orthostatic Hypotension**¹ (Dizziness upon standing)
- **Renal Failure**¹
- **Spinal Stenosis**¹
- **Peripheral Sensory-motor Neuropathy**¹
- **Bilateral Carpal Tunnel Syndrome**¹
- **Chronic GI Distress**¹
- **Autonomic Neuropathy**¹

If you have tingling or numbness in your hands and feet, your doctor may want you to get a biopsy (a procedure to remove a piece of tissue or a sample of cells from your body so that it can be analyzed in a laboratory). If a biopsy confirms amyloid deposits, you should discuss genetic testing for hATTR amyloidosis with your doctor.

You should also discuss a PYP diagnostic test with your doctor, a procedure involving a scan and blood work performed to confirm or determine if the disease has impacted the heart.²

Managing the symptoms of hATTR amyloidosis is an ongoing process because the condition can get worse each day. While there are currently no approved treatments specifically for hATTR amyloidosis in the U.S., your doctor may prescribe medicines to treat the symptoms and the condition’s impact on your daily life.³