Hereditary ATTR amyloidosis: an inherited condition

hATTR amyloidosis is an autosomal dominant disease caused by a mutation in the transthyretin (TTR) gene that results in misfolded TTR proteins accumulating as amyloid fibrils in multiple sites including the nerves, heart, and GI tract.

Formation of amyloid fibrils

The disease affects multiple organs, resulting in varying symptoms

Because amyloid fibrils are deposited in tissues throughout the body, including the nerves, heart, and GI tract, individuals with hATTR amyloidosis can present across a spectrum that includes sensory and motor, cardiac, and autonomic symptoms.

Constellation of possible signs and symptoms of hATTR amyloidosis


The most common signs and symptoms of hATTR amyloidosis include peripheral neuropathy, symptoms of cardiac involvement, early signs of autonomic dysfunction such as GI symptoms (diarrhea, constipation) or sexual dysfunction, unexplained weight loss, carpal tunnel syndrome, and vitreous opacities.7
Confirming an hATTR amyloidosis diagnosis

When you suspect hATTR amyloidosis, the diagnostic process may include:\(^9,11\):
  - Cardiac and/or neurologic examination
  - Genetic testing
  - Tissue biopsy
  - Identification of the amyloid protein

Learn more about red-flag symptoms, diagnostic tools, and differential diagnosis at www.hATTRamyloidosis.com.

Alnylam-sponsored, third-party genetic screening and counseling programs for hATTR amyloidosis made available at no charge

Genetic testing and counseling may help to:
  - Identify risk of disease for patients and their family members
  - Shorten the time to diagnosis and prevent misdiagnoses
  - Determine if patients are eligible to participate in clinical trials
  - Provide information about support resources such as patient advocacy organizations

The Alnylam Act\(^\circ\) program was developed to reduce barriers to genetic testing and counseling to help people make more informed decisions about their health. 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 who use this program have no obligation to recommend, purchase, order, prescribe, promote, administer, use or support any Alnylam product.

Visit www.AlnylamAct.com to learn more about Alnylam’s genetic screening available at no charge.