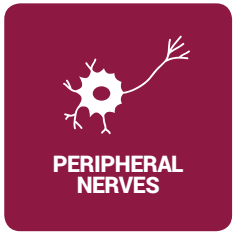
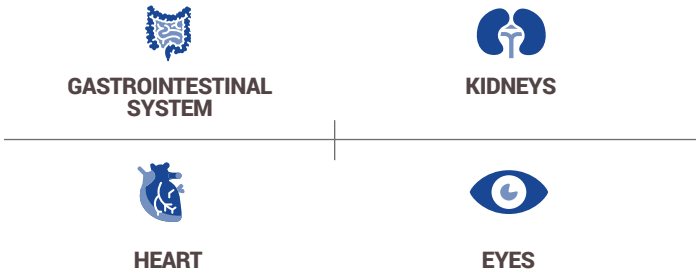
HEREDITARY TRANSTHYRETIN (hATTR) AMYLOIDOSIS //INFOGRAPHIC

What is hATTR amyloidosis?

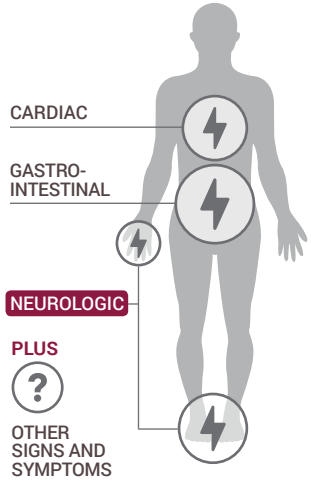
Hereditary transthyretin amyloidosis is a rare, progressive and debilitating genetic disease caused by a gene change or mutation in the transthyretin (TTR) protein.[1,2] This structural change causes TTR to misfold, clump together and accumulate in various tissues and organs throughout the body, including[2,3]:





PATIENT POPULATION

hATTR amyloidosis is estimated to affect **50,000 people worldwide**[3] with more than **130 different TTR gene mutations** identified[4]



Although some mutations are more predominantly associated with polyneuropathy (damage to the nervous system) or cardiomyopathy (damage to the heart muscles), symptoms can differ among affected family members and many patients have mixed clinical phenotypes including **neurologic**, **cardiac**, **gastrointestinal** (GI) and **other signs and symptoms**.[2,3,5,6]

DIAGNOSIS

Because patients often present with seemingly unrelated symptoms that resemble other, more common conditions, it can often **take more than 4 years and visits with 5 or more doctors** across different specialties to receive an accurate diagnosis.[7]

DISEASE PROGRESSION

As the disease progresses, symptoms of hATTR amyloidosis increase in severity and may eventually rob patients of sensory, motor and autonomic functions—affecting their ability to do daily activities, resulting in a major impact on their quality of life.

SYMPTOMS[2,3]

- ✓ **Numbness or tingling of fingers and toes**
- ✓ **Severe constipation and diarrhea**
- ✓ **Shortness of breath**
- ✓ **Bilateral carpal tunnel**
- ✓ **Spinal stenosis**
- ✓ **Sexual dysfunction**

References: 1. Adams D, Amitay O, Coelho T. Patients with hereditary ATTR amyloidosis experience an increasing burden of illness as the disease progresses.*Orphanet J Rare Dis*. 2015;10(suppl 1):P58. **2.** Conceição I, González-Duarte A, Obici L, et al. "Red-flag" symptom clusters in transthyretin familial amyloid polyneuropathy. *J Peripher Nerv Syst*. 2016;21(1):5-9. **3.** Gertz MA. Hereditary ATTR amyloidosis: burden of illness and diagnostic challenges. *Am J Manag Care*. 2017;23(suppl 7):S107-S112. **4.** Ueda, Mitsuharu, and Yukio Ando. Recent Advances in Transthyretin Amyloidosis Therapy. *Translational Neurodegeneration* 3 (2014): 19. PMC. Web. 26 Sept. 2018. **5.** Coelho T, Maurer MS, Suhr OB. THAOS—The Transthyretin Amyloidosis Outcomes Survey: initial report on clinical manifestations in patients with hereditary and wild-type transthyretin amyloidosis. *Curr Med Res Opin*. 2013;29(1):63-76. **6.** Hawkins PN, Ando Y, Dispenzeri A, Gonzalez-Duarte A, Adams D, Suhr OB. Evolving landscape in the management of transthyretin amyloidosis. *Ann Med*. 2015;47(8):625-638. **7.** Amyloidosis Foundation and Amyloidosis Support Groups. Understanding the patient voice in hereditary transthyretin-mediated amyloidosis (ATTR amyloidosis). Last Accessed May 25, 2018. http://amyloidosissupport.org/support_groups/fam_isabell_attr.pdf

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