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. This structural change causes TTR to misfold, clump together and accumulate in various tissues and organs throughout the body, including:

**PATIENT POPULATION**

hATTR amyloidosis is estimated to affect 50,000 people worldwide with more than 130 different TTR gene mutations identified.

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.

**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.

**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**

- 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.