Understanding Hereditary ATTR Amyloidosis (hATTR amyloidosis)

hATTR amyloidosis is an inherited, progressive disease caused by a genetic mutation that results in the buildup of misfolded transthyretin (TTR) protein. This results in the formation of amyloid deposits in the heart, nerves, and GI tract. hATTR amyloidosis affects approximately 50,000 people worldwide. Treatment options for people with this disease are limited and many people remain undiagnosed or misdiagnosed.

CAUSE

People with hATTR amyloidosis have a genetic mutation that prevents TTR protein, which is made in the liver, from performing its normal function. Instead, TTR misfolds and accumulates as amyloid deposits in the body—in the heart, nerves, and GI tract, as well as other organs—causing symptoms.

SYMPTOMS

The symptoms of hATTR amyloidosis vary from person to person, depending on which organs or tissues are affected. As the disease progresses, symptoms may worsen (e.g., numbness or tingling of the feet can result in the need to walk with an aid or use a wheelchair) and can lead to significant disability, decreased quality of life, and, in many instances, a shortened lifespan.

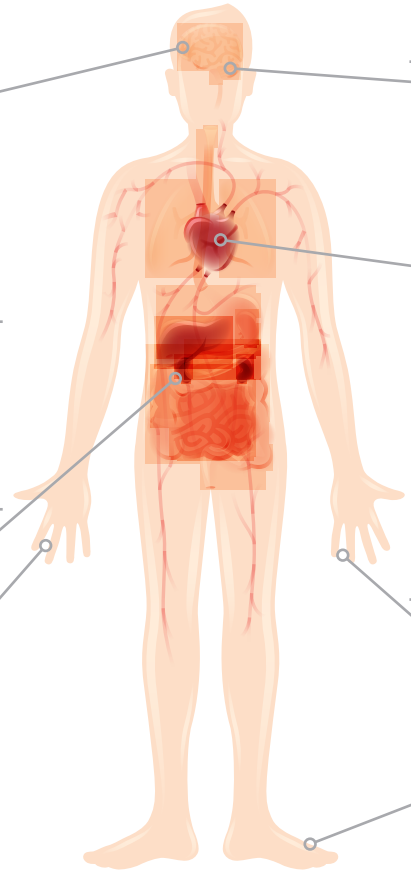
The Varying Symptoms of hATTR Amyloidosis

- Progressive dementia
- Headache
- Loss of movement control
- Seizures
- Weakness
- Stroke-like episodes

- Nausea and vomiting
- Feeling full early
- Diarrhea
- Severe constipation
- Alternating episodes of diarrhea and constipation
- Unintentional weight loss

- Excess protein in urine
- Kidney failure

- Carpal tunnel syndrome



- Glaucoma
- Blurred or spotty vision
- Abnormalities of the pupil or blood vessels on the white of the eye

- Irregular heartbeat
- Heart failure
- Leaky heart valves

- Decreased blood pressure upon standing
- Urinary tract infections
- Sexual dysfunction
- Abnormal sweating

- Swelling (edema of the legs and feet)
- Altered sensation and/or mobility in the arms, hands, legs and feet
  - Numbness/tingling
  - Weakness
  - Pain

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DIAGNOSIS

People affected with hATTR amyloidosis need an early and accurate diagnosis due to the potential for rapid disease progression. Misdiagnosis of hATTR amyloidosis is common because of the number of symptoms that overlap with those of many other diseases, which may result in potentially ineffective or harmful treatment.

Procedures commonly used to confirm the diagnosis of hATTR amyloidosis include blood tests, organ biopsies, and tissue biopsies. Genetic testing can be used to confirm a mutation in the TTR gene. Once a TTR gene mutation is identified, family members of an affected individual can use this information to help determine their own risk. hATTR amyloidosis is passed down when one parent carries the mutation, giving children a 50% chance of inheriting that mutation. Some people with a hATTR gene mutation may not experience symptoms. Genetic counseling can be helpful for individuals at risk of developing hATTR amyloidosis (i.e., individuals with family members carrying the mutation).

TREATMENTS

There are no medicines approved by the FDA for hATTR amyloidosis. Affected individuals may work with an amyloidosis specialist and a coordinated health care team to determine an individualized disease management plan to manage symptoms. Liver transplants may be an option for some individuals. Therapies are in development for the treatment of hATTR amyloidosis.

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