Transthyretin Amyloid Cardiomyopathy (ATTR-CM)

An Underdiagnosed and Life-Threatening Disease

ATTR-CM is a rare, underdiagnosed condition that is associated with progressive heart failure.¹

ATTR-CM: A Presentation of Transthyretin Amyloidosis

Transthyretin amyloidosis is a rare, progressive disease characterized by the buildup of abnormal deposits of misfolded proteins called amyloid (amyloidosis) in the body’s organs and tissues.²,³

Transthyretin amyloidosis can impact numerous areas in the body, including the peripheral nervous system, and organs such as the heart, kidney, gastrointestinal tract, and eyes.²

ATTR-CM is a presentation of the disease that affects the heart and is defined by restrictive cardiomyopathy and progressive heart failure.²

Causes and Symptoms of ATTR-CM

ATTR-CM is caused when transthyretin, a transport protein that naturally circulates in the blood, becomes unstable and misfolds. The misfolded protein can build up in the heart as amyloid fibrils, which causes the heart muscle to become stiff, eventually resulting in heart failure.³,⁴

Symptoms include shortness of breath, fatigue, and peripheral (e.g., ankle) edema. Often the disease is diagnosed only after symptoms have become severe.³
Two Sub-types of ATTR-CM

**HEREDITARY, ALSO KNOWN AS VARIANT**

The inherited form of ATTR-CM, is caused by a mutation in the transthyretin gene, which causes the transthyretin protein to be unstable and misfold. Can occur in people as early as their 50s and 60s. V122I is the most common mutation and in the United States is seen predominately in African Americans or people of African descent.

**WILD TYPE (ATTRwt)**

In wild-type ATTR-CM, there is no mutation and the disease is not inherited. Instead, it is associated with aging and the protein becomes unstable and misfolds. A majority of patients with ATTR-CM have wild type, which usually affects men after age 60.

Challenges and Impact

- In ATTR-CM, receiving a diagnosis has historically been difficult because disease awareness is low among health care professionals and patients often present with symptoms similar to more common causes of heart failure.
- The prevalence of ATTR-CM is presently unknown, but it is believed that less than 1% of people with the disease are diagnosed.
- The average life expectancy for people with ATTR-CM is approximately 2 to 6 years from diagnosis.

Learn More & Find Support

**Amyloidosis Research Consortium:**
http://www.arci.org

**Amyloidosis Foundation:**
www.amyloidosissupport.org