Transthyretin amyloid cardiomyopathy (ATTR-CM)

A rare, underdiagnosed, life-threatening disease associated with progressive heart failure

**ATTR-CM: A Presentation of Transthyretin Amyloidosis**

*Transthyretin amyloidosis* is a rare, progressive disease characterized by the abnormal buildup of amyloid deposits composed of misfolded transthyretin protein in the body’s organs and tissues; this build up is called amyloidosis.

ATTR amyloidosis can impact numerous organs and tissues in the body, including the peripheral nervous system, and organs such as the heart, kidneys, gastrointestinal tract, and eyes.

**ATTR-CM** is a presentation of the disease that affects the heart and leads to restrictive cardiomyopathy and progressive heart failure.

**Causes and Symptoms of ATTR-CM**

ATTR-CM occurs when when transthyretin, a transport protein that naturally circulates in the blood, becomes unstable, and dissociates into monomers, which misfold. The misfolded protein aggregates into amyloid fibrils which build up in the heart and causes the heart muscle to become stiff, eventually resulting in heart failure.

Symptoms can include shortness of breath, fatigue, and swelling of the ankles. Often the disease is diagnosed only after symptoms have become severe.
Two Sub-types of ATTR-CM

HEREDITARY, also known as variant

The hereditary form of ATTR-CM is inherited and caused by a mutation in the transthyretin gene, which results in an unstable transthyretin protein that dissociates and misfolds, forming amyloid. Can occur in people as early as their 50s and 60s.

Wild Type

The wild-type form of ATTR-CM, which is thought to be more common, usually affects men after age 60.

Challenges and Impact

In ATTR-CM, receiving a correct diagnosis has historically been difficult because disease awareness is low among health care professionals, and misdiagnosis is common because patients often present with symptoms similar to more common causes of heart failure.

ATTR-CM is significantly under or misdiagnosed, making it difficult to characterize worldwide prevalence. It is believed that only 1-2% of people with the disease are diagnosed. Education is important to help recognize symptoms and improve diagnosis.

Without treatment, the average life expectancy for people with ATTR-CM is approximately 2.3-3.5 years from diagnosis.

Learn More & Find Support

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

Amyloidosis Foundation:
www.amyloidosisresearchfoundation.org

Amyloidosis Alliance:
https://www.amyloidosisalliance.org/

Amyloidosis Support Groups:
http://amyloidosissupport.org

MacKenzie’s Mission:
https://mm713.org/

Amyloidosis Patient Information Site:
https://www.amyloidosis.org.uk/about-amyloidosis/introduction-to-attr-amyloidosis/

8. 2018 Internal Analysis, Data on File Pfizer Inc.