

here to answer your questions. ■

Transthyretin Amyloid Cardiomyopathy (ATTR-CM) Frequently Asked Questions



Q: What is transthyretin amyloidosis?

A: Amyloidosis is a disease characterized by the buildup of abnormal deposits of a protein in the body's organs and tissues; this build-up is called amyloid (amyloidosis). Transthyretin amyloidosis occurs when the precursor protein is transthyretin, a transport protein which most frequently impacts the peripheral nervous system and organs such as the heart, kidney, GI tract, and eyes.¹

Q: What is ATTR-CM?

A: ATTR-CM is a rare, progressive form of transthyretin amyloidosis characterized by the buildup of abnormal deposits of misfolded proteins called amyloid in the body's organs and tissues. It is a presentation of transthyretin amyloidosis, that primarily affects the heart and is defined by restrictive cardiomyopathy and progressive heart failure.^{1,2}

Q: What causes ATTR-CM?

A: It 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, resulting in heart failure.^{1,2}



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Q: What is amyloid?

A: Amyloid refers to the abnormal fibrous deposits which are insoluble and composed of aggregates of misfolded proteins.³

Q: What are the symptoms of ATTR-CM?

A: People with ATTR-CM often present with symptoms similar to more common types of heart failure, which can lead to a misdiagnosis. These symptoms include shortness of breath, fainting, fatigue, and peripheral (e.g., ankle) edema. Often the disease is diagnosed only after symptoms have become severe.^{1,2}

Q: Is ATTR-CM inherited?

A: There is a form that is hereditary, also known as variant and caused by a mutation in the transthyretin gene. There is also a non-hereditary form, known as wild type (ATTRwt), that is associated with aging. In both types, the transthyretin protein becomes unstable and then misfolds, accumulating as amyloid in the heart.^{1,3}

Q: How is ATTR-CM diagnosed?

A: The signs and symptoms of ATTR-CM are similar to those of patients presenting with heart failure, including dyspnea, fatigue, and edema. However, ATTR-CM should be suspected when patients experience heart failure with preserved ejection fraction, and/or left ventricular hypertrophy with abnormal ratio between LV thickness and QRS voltage and a poor response to heart failure medicines. Non-invasive diagnostic tests such as echocardiogram, electrocardiogram, and cardiac MRI can help to identify patients at risk for disease and suggest the presence of amyloid in the heart. A final diagnosis to confirm ATTR-CM is made with a combination of genetic testing and pyrophosphate scintigraphy—an emerging diagnostic technique—or cardiac biopsy.^{2,5,6}

Q: Who is typically diagnosed with ATTR-CM?

A: ATTR-CM typically occurs during adulthood. Mutated transthyretin amyloidosis can occur in people as early as their 50s and 60s, while the wild-type transthyretin amyloidosis is believed to account for the vast majority of diagnosed patients and usually occurs or presents after age 60.^{1,4,7}

Q: How common is ATTR-CM?

A: The prevalence of ATTR-CM is presently unknown. It is believed that less than 1% of people with ATTR-CM are diagnosed.⁸

Q: Why is the diagnosis rate of ATTR-CM low?

A: Diagnosing ATTR-CM is challenging, as disease awareness is low and people often present with symptoms similar to more common types of heart failure. Because of this, education is vital to increase awareness and understanding of transthyretin amyloid cardiomyopathy. This may potentially help to identify people with the disease earlier.⁹

Q: What treatment options are available for ATTR-CM?

A: Currently, there are no approved pharmacologic treatment options for ATTR-CM. Management options for transthyretin amyloid cardiomyopathy include symptom management and in select cases, heart and/or liver transplants.¹⁰

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