Transthyretin amyloid cardiomyopathy (ATTR-CM)

**ATTR-CM: An Underdiagnosed Cause of Heart Failure**

*Transthyretin amyloid cardiomyopathy,* or *ATTR-CM,* is a rare and life-threatening condition that affects the heart and is associated with heart failure.¹²

It is the result of misfolding proteins that become unstable, resulting in the creation of amyloid fibrils which build up in the heart and other parts of the body.¹²

The buildup of misfolded proteins causes the heart muscle to stiffen over time, eventually leading to heart failure.¹²

**Signs and Symptoms of ATTR-CM**

*Symptoms* often mimic other more common types of heart failure and can include shortness of breath, fatigue, and swelling of the ankles, but may also include other symptoms related to buildup of amyloid fibrils throughout the body, such as carpal tunnel syndrome and peripheral neuropathy.

Often the disease is diagnosed only after symptoms have become severe. Patients should talk to their cardiologist about ATTR-CM if they have heart failure and experience any of these signs and symptoms.³¹¹

**Signs and Symptoms**

- **FATIGUE**
- **SHORTNESS OF BREATH**
- **ARRHYTHMIA**
  - Irregular heartbeat
- **BILATERAL CARPAL TUNNEL SYNDROME**
  - Numbness, tingling, and pain in the fingers
- **LUMBAR SPINAL STENOSIS**
  - Pain or numbness in the lower back and legs due to narrowing of lower spine
- **PERIPHERAL NEUROPATHY**
  - Decreased or strange tingling sensation or pain in feet or toes
- **GASTROINTESTINAL ISSUES**
  - Such as diarrhea, constipation, nausea or feeling full quickly
- **HIP AND/OR KNEE REPLACEMENT**
- **PERIPHERAL EDEMA**
  - Swelling in the lower legs
- **BICEPS TENDON RUPTURE**
  - Not related to trauma
- **OCULAR MANIFESTATIONS**
  - (ie, glaucoma)
Two Sub-types of ATTR-CM

Hereditary (hATTR-CM), also known as variant

Hereditary ATTR-CM occurs due to a mutation in the transthyretin gene. It can occur in people as early as their 50s and 60s. Not all people with a TTR mutation will develop hATTR-CM.

Wild-type (wtATTR-CM)

The wild-type form of ATTR-CM is associated with aging and is thought to be the most common form of ATTR-CM.

Challenges and Impact

In ATTR-CM, receiving a correct diagnosis has historically been difficult because disease awareness is low among healthcare professionals, and misdiagnosis is common because patients often present with symptoms similar to more common causes of heart failure.15

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.15 Education is important to help recognize symptoms and improve diagnosis.14

Without treatment, the average life expectancy for people with ATTR-CM is approximately 2—3.5 years from diagnosis.16,17

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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/

One Amyloidosis Voice: https://www.oneamyloidosisvoice.com/