

Living with *Transthyretin Amyloid Cardiomyopathy (ATTR-CM)*

A DISCUSSION GUIDE FOR PATIENTS AND CAREGIVERS

WHAT IS ATTR-CM?

ATTR-CM, also known as transthyretin amyloid cardiomyopathy, is a serious and severe condition that affects the heart muscle. It develops when a protein called **transthyretin (TTR)** changes its shape and forms clumps (amyloid deposits) in the heart – and sometimes in the nerves or other parts of the body. This process is called **amyloidosis**.

Over time, these deposits can make the walls of the heart become stiff. This can cause a type of heart disease called **restrictive cardiomyopathy**, which makes it difficult for the heart to pump blood properly. If left untreated, ATTR-CM can lead to **heart failure**.

TYPES OF ATTR-CM



HEREDITARY (hATTR-CM)

This type is caused by a change in a gene, known as genetic mutation, which affects how the TTR protein works. It is **inherited** and can be passed down through families – **if one parent has hATTR-CM, there is a 50% chance their child could inherit it**. It affects both men and women, usually between ages 50 – 60.



WILD-TYPE (wtATTR-CM)

This form is **not inherited** and doesn't result from a genetic mutation. Although the exact cause is unknown, it is thought to be linked with aging and is most often seen in men over 60 years old. As people are living longer, this type is becoming more common. In fact, compared to men, the proportion of women diagnosed with the wild-type is higher after 80 years old, as they tend to live longer than men.



UNDER-DIAGNOSED:

MANY PEOPLE LIVING
WITH ATTR-CM REMAIN
undiagnosed



OVERLOOKED:

ATTR-CM CAN BE
*missed
altogether*



MISDIAGNOSED:

BECAUSE ITS SYMPTOMS
ARE SIMILAR TO OTHER
HEART CONDITIONS,
ATTR-CM CAN EASILY BE
*mistaken for
something else*

SYMPTOMS OF ATTR-CM

The symptoms of ATTR-CM are often similar to those of other heart conditions, which can delay diagnosis. Some people may have no signs or only mild ones and go unnoticed for years, until the disease reaches a more advanced stage.

The condition can affect each person differently. Some common symptoms may include:



SHORTNESS OF BREATH
(most common)



FATIGUE
excessive tiredness



THICKENED TONGUE



GASTROINTESTINAL ISSUES
(e.g., diarrhoea)



CARPAL TUNNEL SYNDROME
(a condition that causes pressure on a nerve in the wrist)



NUMBNESS
strange tingling sensation, or pain in the toes or feet



SWELLING
in the lower legs

If your symptoms are getting worse even while taking heart failure medication, or if you are having adverse effects, talk to your healthcare provider.

DIAGNOSING ATTR-CM

Because ATTR-CM shares symptoms with other heart conditions — and sometimes causes few or no noticeable symptoms at all — it can often be overlooked, which is why reaching a diagnosis can take time. The good news is that with improved awareness and testing, it is possible to get a clear diagnosis and start appropriate treatment.

To confirm a diagnosis, your healthcare provider may perform a **physical exam** to assess your overall health, review your **medical records** and ask about your **family's health**. Additional tests to confirm a diagnosis of ATTR-CM may include:

- ♥ **BLOOD TESTS**
To check the levels of substances normally in the blood and to look for markers of amyloidosis or heart muscle damage.
- ♥ **URINE TESTS**
To help assess kidney function and rule out other forms of amyloidosis.
- ♥ **GENETIC TESTING (blood or saliva)**
Helps determine if ATTR-CM is hereditary or wild-type and can guide the decision whether other family members should consider getting tested too.
- ♥ **TECHNETIUM-LABELLED CARDIAC SCINTIGRAPHY (99m Tc-PYP scan)**
Uses a radioactive tracer (99mTc-PYP) that accumulates in amyloid deposits in the heart. While this is a similar technology to a bone scan, it does not involve the bones.
- ♥ **TISSUE BIOPSY**
Involves examining a small sample of heart tissue under a microscope to confirm the presence of amyloid deposits.

*Additional tests may be needed to confirm a diagnosis of cardiomyopathy. Some patients might also need to use portable devices like **Holter monitors** or **event monitors**. These devices are typically worn for 24 to 48 hours to continuously record the heart's electrical activity (EKG) during normal daily activities.*

EARLY DETECTION MATTERS:

GETTING THE

right diagnosis early

CAN IMPROVE THE LIFE FOR THOSE LIVING WITH ATTR-CM

TREATING AND MANAGING ATTR-CM

While there is currently no cure for ATTR-CM, treatments can help slow the disease down by reducing or stopping the buildup of harmful proteins in the heart. They can also improve symptoms, support your heart's function, and help manage complications like heart failure and heart rhythm issues.

Your medical team — which may include cardiologists, radiologists, neurologists, and other specialists — will work with you to create a treatment plan tailored to the type and severity of your condition.



HEALTHY LIFESTYLE: Adopting healthy habits like **eating nutritious foods**, maintaining a **proper weight**, engaging in **regular physical activity**, **controlling blood pressure and blood sugar levels**, **not smoking**, **limiting salt intake** and **reducing stress**, can improve your overall well-being and longevity.



MEDICATIONS: Certain medicines can slow the disease from getting worse by stopping the protein from building up in the body. Others help relieve cardiomyopathy and/or heart failure symptoms like swelling from fluid buildup, irregular heartbeat, or high blood pressure — so you can feel better and stay more active. People with a restrictive cardiomyopathy are also at increased risk for developing blood clots in their heart chambers, so in this situation anti-blood clotting drugs may be prescribed.



DEVICES: These include **pacemakers** for controlling heart rate and rhythm, **implantable cardiac defibrillators (ICDs)** that deliver a shock to the heart to correct life-threatening arrhythmia, and **left ventricular assist devices (LVADs)** to help pump blood from the heart's left lower chamber (ventricle) to the rest of the body.



TRANSPLANT SURGERY: In severe cases, when all other treatments have failed to adequately improve the heart's condition, a **heart transplant** may be the only option. In some cases of hATTR-CM, a **liver transplant** may be considered to stop the production of the abnormal TTR protein, however this option is only used for the hereditary form.

QUESTIONS TO ASK YOUR HEALTHCARE PROVIDER

- ✓ Could ATTR-CM be causing my symptoms?
- ✓ I have heart failure, could this be caused by ATTR-CM?
- ✓ What type of ATTR-CM am I diagnosed with and what complications could arise from my condition?
- ✓ How severe is my ATTR-CM? Are other organs affected?
- ✓ Should I have genetic testing? Should my family members?
- ✓ What are my treatment options, and what are their risks and benefits?
- ✓ Are there any medications I should avoid?
- ✓ What happens if I don't get treatment?
- ✓ What lifestyle changes should I make?
- ✓ How should I monitor my condition over time?
- ✓ How often should I have routine visits with you?
- ✓ When should I seek emergency care?
- ✓ How can I connect with other individuals facing ATTR-CM for support and shared experiences?



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