



Transthyretin Amyloid Cardiomyopathy (ATTR-CM)

Fast Facts:

- Rare heart disease caused by buildup of a faulty protein.
- If untreated, can get worse over time leading to heart failure and death.
- With new testing and treatments, more cases are being diagnosed and treated.

What is ATTR-CM?

- Disease of the heart muscle caused by a protein (transthyretin) normally found in the bloodstream.
- Two types:
 - Hereditary (mutation in the genes)
 - Wild type (normal genes).

What causes ATTR-CM?

- In ATTR-CM, transthyretin is misshaped and can form clumps called amyloid. These clumps can build up in the heart, nerves and other organs.
- When amyloid builds up in the heart, the walls can become stiff, making it harder for the heart to relax and fill with blood. As the disease gets worse, the heart becomes unable to pump blood out of the heart, leading to heart failure.

How is ATTR-CM Diagnosed?

- Nuclear scan of the heart called cardiac PYP scan, where a radioactive dye is injected before the scan is most commonly used now to diagnose ATTR-CM.
- Rarely there are cases where an actual heart tissue biopsy is required for diagnosis.
- Genetic testing is routinely offered to detect genetic change in the transthyretin gene.

How is ATTR-CM Treated?

- Currently, a transthyretin stabilizer called tafamidis (Vyndamax® or Vyndaqel®) is approved for ATTR-CM. By stabilizing the misshaped transthyretin protein, there will be less clumps forming and building up in the heart and other organs, leading to better long-term outcome.
- For hereditary ATTR that is also associated with nerve damages, transthyretin gene silencer medicines like patisiran (Onpattro®), vutrisiran (Amvuttra®) or eplontersen (Wainua®) can reduce nerve symptoms by minimizing the production of misshaped transthyretin altogether.

Points to Remember:

- Early treatment can slow the worsening of the disease and improve survival.
- A lot of ongoing clinical trials can also be considered as treatment options.

For Additional Information:

- Amyloidosis Support Groups - <https://www.amyloidosisupport.org/>
- Amyloidosis Foundation - <https://www.amyloidosis.org/>