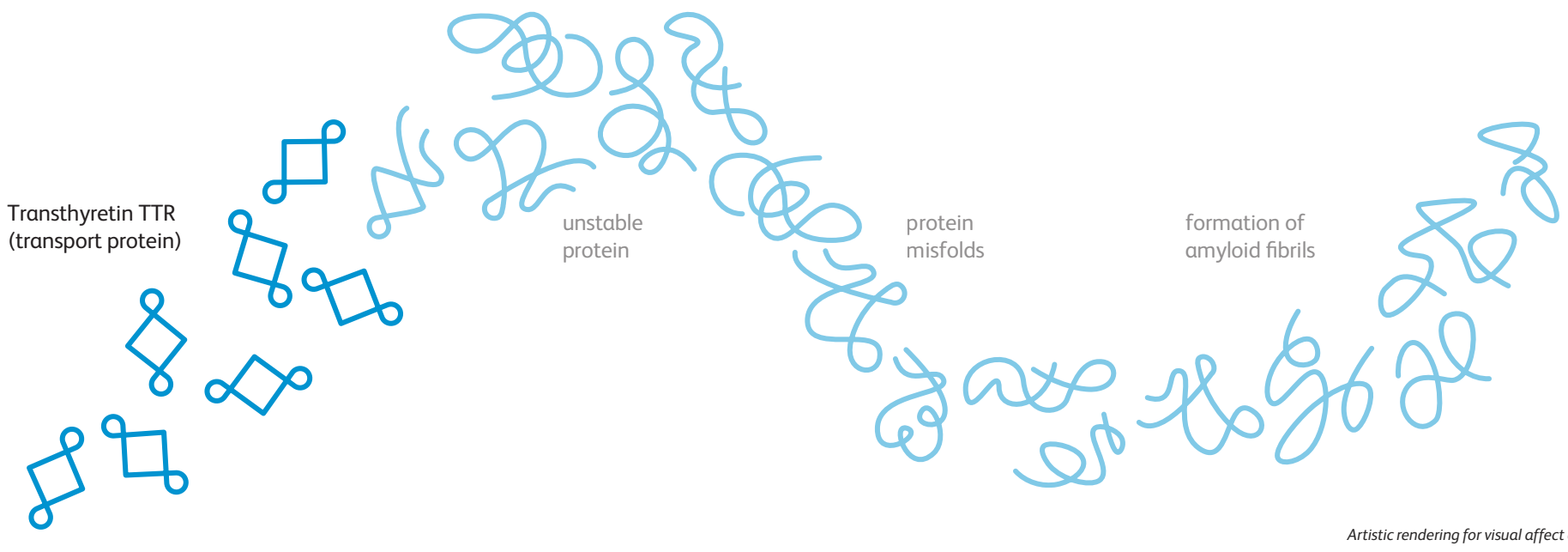
Transthyretin Amyloid Cardiomyopathy (ATTR-CM)

The Facts

Amyloidosis refers to a group of deposition diseases in which proteins with unstable structures **misfold and aggregate into amyloid fibrils** that deposit in different organs and tissues.[1,2]

ATTR-CM is a **serious and underdiagnosed** type of cardiac amyloidosis that is associated with heart failure. The unstable, misfolded proteins can build up in the heart and other parts of the body, causing the heart muscle to stiffen over time, eventually leading to heart failure.[1,2]



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Transthyretin, a protein produced primarily by the liver, dissociates into monomers, which misfold and aggregate into amyloid fibrils. When the fibrils deposit in the heart, this can cause ATTR-CM.

There are two subtypes of ATTR-CM:

Hereditary, or variant ATTR-CM (hATTR) is inherited from a relative and is caused by a mutation in the gene that produces the transthyretin protein. It affects both men and women, with symptom onset occurring in people as early as their 50s or 60s. In the U.S., the most common type (V122I) is found almost exclusively in individuals of African ancestory.[1-6]

Wild-type ATTR-CM (wtATTR) is associated with aging and is thought to be the most common form of ATTR-CM. This form most often affects white men over the age of 60 and is not caused by a genetic mutation.[1-6]

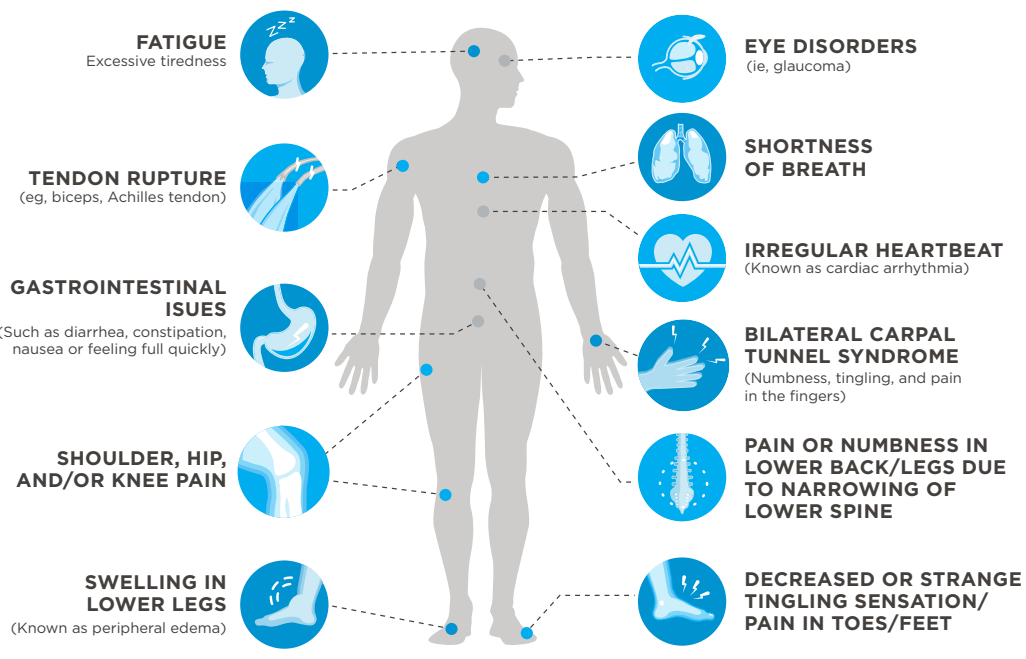
A Difficult Road to Diagnosis

ATTR-CM is believed to be **rare** and **significantly under or misdiagnosed** though the actual number of people with the disease is currently unknown.[8]

Awareness of ATTR-CM is low within the physician community, and as a result, continues to be significantly underdiagnosed. Often ATTR-CM is diagnosed only after symptoms have become severe.[8]

Once diagnosed, the average life expectancy for people with ATTR-CM is approximately 2 to 3.5 years if left untreated.[7]

Signs and Symptoms May Include:[1,11-19]



How is ATTR-CM diagnosed?

ATTR-CM is suspected based on the signs and symptoms presented, as well as results from ECHO, ECG and/or Cardiac MRI.[9,10]

Once another form of cardiac amyloidosis (light chain amyloidosis) is ruled out by blood and urine analysis, the following tests can contribute to the diagnosis of ATTR-CM following clinical suspicion:[9,10]

NUCLEAR SCINTIGRAPHY (NON-INVASIVE DIAGNOSTIC TEST TO CONFIRM PRESENCE OF AMYLOID IN THE HEART)

CARDIAC BIOPSY (AN INVASIVE TEST TO CONFIRM THE PRESENCE OF TTR AMYLOID DEPOSITS)

GENETIC TESTING USED TO DETERMINE IF THE DISEASE IS HEREDITARY DUE TO A MUTATION IN THE TTR GENE

Treatment Challenges

Until recently, there were no medicines approved for the treatment of ATTR-CM. Historically, management options were limited to symptom management, and in rare cases, heart or heart and liver transplant.[2]

Patients should talk to their cardiologist about ATTR-CM if they have heart failure and experience any of the signs or symptoms described above.

Education, awareness, and treatment are critical to improving the diagnosis and care of people affected by ATTR-CM.

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