

Uncover the ATTR DIAGNOSIS PROCESS

Understand the signs, symptoms,
and tests needed for an accurate
diagnosis of ATTR amyloidosis (ATTR).

➞ Signs & Symptoms and Common Misdiagnoses

➞ Assessments and Testing

➞ Genetic Testing and Counseling

➞ Next Steps After a Diagnosis

Recognize the signs and symptoms

ATTR is an underdiagnosed disease with symptoms that can worsen over time and affect multiple parts of the body, including the **heart, nerves, bodily functions, muscles, joints, and tendons.**

Monitoring your symptoms, even if they seem unrelated, is a key piece of information that your doctor uses to make a diagnosis.

Reveal more with the Symptoms Tracker & Checklist at ATTRrevealed.com/tracker

Getting the right diagnosis is key

Seeking **an accurate diagnosis as soon as possible is crucial** to taking the next steps in your journey with ATTR.

Misdiagnosis or delays in diagnosing ATTR are common because the varying symptoms can be mistaken for other conditions, including:

- **Hypertensive heart disease:** damage to the heart caused by high blood pressure
- **Aortic stenosis:** narrowing of the aortic heart valve
- **Hypertrophic cardiomyopathy:** disease that thickens the wall of the left ventricle of the heart and impairs heart function
- **Chronic inflammatory demyelinating polyneuropathy (CIDP):** autoimmune disorder in which the body attacks its own nerves
- **Idiopathic neuropathy:** damage to the nerves with an unclear cause
- **Diabetic neuropathy:** nerve damage caused by diabetes

Tell your doctor about all the symptoms you are experiencing, even if they don't apply to your doctor's specialty. Having open and honest conversations about your health with your doctor can help you get a diagnosis sooner.

Preliminary tests

If your doctor thinks you may have ATTR, they may conduct assessments of your heart and nerve function, or order imaging studies or laboratory tests to **determine the cause of your symptoms, including:**

TESTS THAT MEASURE HOW WELL YOUR HEART IS WORKING:

- **Electrocardiography (ECG/EKG)** can detect heart-related conditions. Electrodes are used to record electrical activity and the rhythm of the heart
- **Echocardiography (Echo)** is a type of imaging that uses ultrasound to visualize the heart structure and measure heart valve function and cardiac muscle strength
- **Cardiac magnetic resonance imaging (CMRI)** is a type of imaging that uses a magnetic field to visualize the heart structure and determine how well blood flows through the heart

TESTS THAT MEASURE HOW WELL YOUR NERVES ARE WORKING:

- **A nerve conduction study (NCS)** can detect nerve damage. It uses electrodes to test the speed at which electrical impulses move through the nerves
- **Electromyography (EMG)** can detect abnormalities in the nerves and muscles by measuring the electrical activity of muscle in response to a nerve's stimulation

Confirmatory tests

If your preliminary test results suggest the possibility of ATTR, your doctor may perform additional tests to **help confirm the diagnosis, such as:**

- **Technetium pyrophosphate scintigraphy (PYP scan)**, which is a type of imaging and a non-surgical way of detecting amyloid deposits in the heart
- **Tissue biopsy** to identify amyloid deposits in the tissues
- **Genetic testing** from DNA samples (blood, cheek swab, or saliva) to detect genetic variants in the transthyretin (TTR) gene

These are not complete lists of tests that can assess symptoms or help confirm a diagnosis related to ATTR. Depending on your symptoms, your doctor may order other tests.



Consider genetic counseling and testing

If you have ATTR, ask your doctor if genetic testing is right for you. It is needed to determine if you have wild-type ATTR (wtATTR) or hereditary ATTR (hATTR) and if other family members should get tested to understand their own risk of developing the disease.

Your doctor may also recommend you work with a genetic counselor who can help you and your family learn more about the genetic testing process and if it is right for you.

Genetic counselors are trained healthcare professionals who can:



Work with people who are considering a genetic test and provide guidance after a test



Educate about genetics, inheritance, and disease risk



Discuss the benefits, limitations, and potential implications of genetic testing



Provide information about resources and support

Alnylam Act[®] Genetic Testing and Counseling Program

Alnylam Pharmaceuticals sponsors no-charge, third-party **genetic testing and counseling** for individuals who may carry **one of the 120 or more gene variants** known to be associated with hATTR.

The Alnylam Act program was created to provide access to genetic testing and counseling to patients as a way to help people make more informed decisions about their health.

- While Alnylam provides financial support for this program, tests and services are performed by independent third parties
- Healthcare professionals must confirm that patients meet certain criteria to use the program
- Alnylam receives de-identified patient data from this program, but at no time does Alnylam receive patient-identifiable information. Alnylam may use healthcare professional contact information for research purposes
- Both genetic testing and genetic counseling are available in the US and Canada
- Healthcare professionals or patients who use this program have no obligation to recommend, purchase, order, prescribe, promote, administer, use, or support any Alnylam product
- No patients, healthcare professionals, or payers, including government payers, are billed for this program

Learn more at www.AlnylamAct.com



Keep the conversation going

If you are diagnosed with ATTR, it is important to **have ongoing conversations with your healthcare team and support network**, such as family and friends. Building a solid support system can relieve some of the potential burden of the disease.



CECE,
living with hATTR

Managing the disease

If you are diagnosed with ATTR, starting treatment right away is key to **managing your symptoms**.

There are treatment options available for patients with certain symptoms and types of ATTR that address the underlying cause of the disease. These work by:

SILENCERS

Reducing the amount of TTR protein made in the body, which can help decrease the formation of amyloid deposits

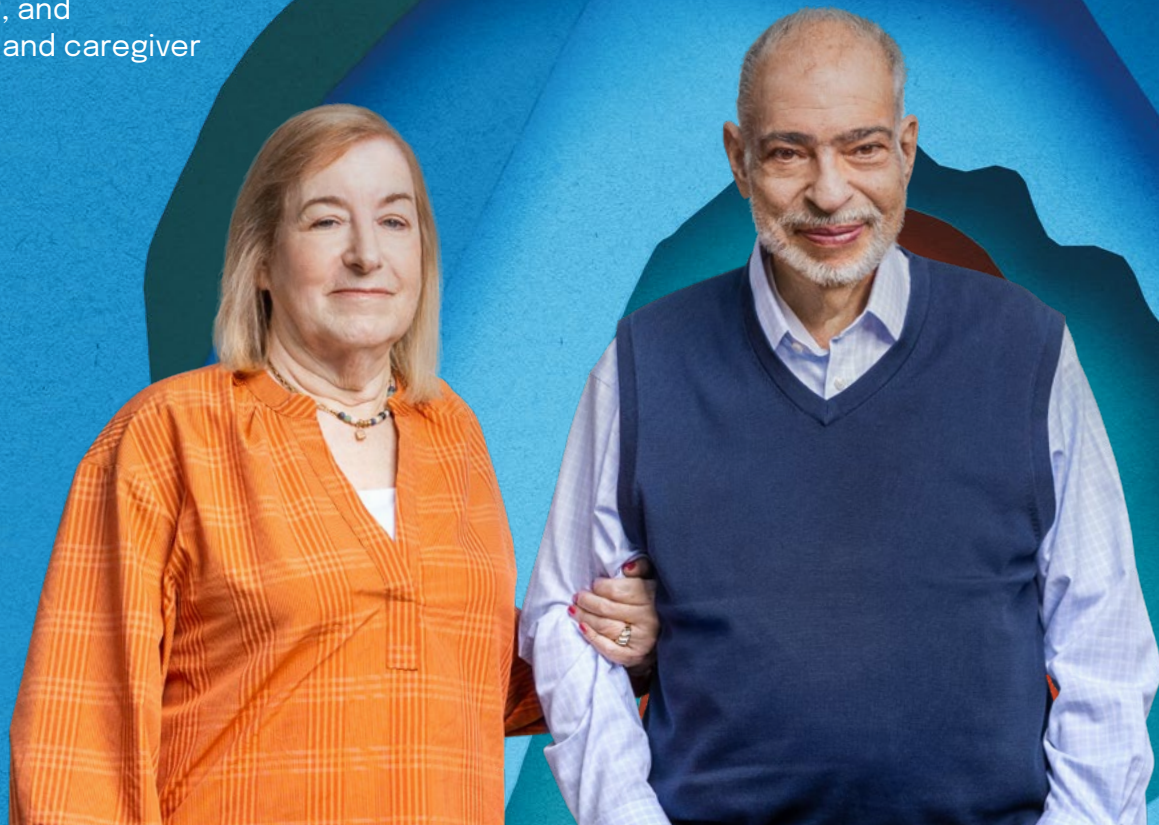
STABILIZERS

Decreasing the amount of TTR proteins that take on an abnormal shape and form amyloid deposits in the body

Since ATTR can worsen quickly over time, it is important to share any new or changing symptoms that you or your loved one are experiencing. Managing the condition is an ongoing process, so it is important to have these conversations with your healthcare team.

Talk to your doctor about what treatment plan may be right for you.

IRV, living with wtATTR, and
REBECCA, his spouse and caregiver



Scan the code or visit www.ATTRrevealed.com
to learn more about ATTR and take advantage
of other resources.



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