

The Role of Genetic Testing in Amyloidosis



Introduction to Genetic Testing

Genetic testing is a process that reveals the presence of specific variations in your genes and can be a pivotal step toward a clear diagnosis, offering a clearer understanding of your health. Exploring your genetic traits through comprehensive genetic testing is crucial for the early detection and effective management of hereditary transthyretin-mediated amyloidosis (hATTR). Timely intervention can significantly impact the progression of this disease.



Navigating Amyloidosis through Genetic Testing

If you're experiencing a mix of conditions or symptoms including:

- Peripheral neuropathy (a condition affecting the nerves outside the brain and spinal cord, often presenting as tingling, numbness, weakness or pain, usually in the hands and feet)
- Heart failure with preserved ejection fraction (HFpEF) accompanied by increased left ventricular wall thickness
- Bilateral carpal tunnel syndrome
- Trigger fingers (a condition characterized by a finger bending, getting stuck and then snapping straight)
- Spinal stenosis
- Sudden biceps tendon rupture
- Prior orthopedic procedures

Genetic testing may be important for identifying the root cause of your symptoms. Especially if you have a family history of hATTR, testing offers a targeted approach for timely diagnosis and personalized care.

Don't ignore the signs—recognizing these symptoms early and taking the steps to identify the cause can pave the way for a more precise and effective management of your health and well-being.



Understanding the Role of Genetic Counseling

As you explore the possibility of genetic testing, genetic counselors can shed light on the procedure, potential outcomes, and associated risks and uncertainties. After the test, they will interpret the results, steer you through the next steps, and offer essential resources and support.

Navigating a challenging health diagnosis can be overwhelming, but genetic testing and counseling can make this process smoother and provide clarity to you and your family. To find additional resources and guidance on navigating a diagnosis and treatment for hATTR, including discussion guides for conversations with family and healthcare providers, visit MyATTRRoadmap.com.