

Understanding Your Negative Transthyretin Amyloidosis (TTR) Genetic Test Result

INFORMATION FOR PATIENTS

Result	NEGATIVE	The result of your genetic testing did not find any mutations (changes) in a gene that causes hereditary transthyretin amyloidosis. Not all patients with amyloidosis have a mutation in this gene. You may have a mutation in a gene that was not included in this test. If someone in your family has a specific mutation in the TTR gene, it is likely that you do not carry that mutation.
Diagnosis	NO CHANGE	This testing does not change your clinical diagnosis. If you have been diagnosed with amyloidosis, that remains the same.
Management Options	PATIENTS WITH AMYLOIDOSIS	Medical management options include: medications, surgery, pacemakers, or liver transplantation. Talk to your healthcare providers about which may be right for you.
Screening Options	PATIENTS WITH A FAMILY HISTORY OF AMYLOIDOSIS, BUT NO PERSONAL SYMPTOMS	Options for screening and early detection include: physical exams, blood testing, cardiac evaluations, or tissue biopsy analysis. Talk to your healthcare providers about whether these options are right for you.
Next Steps	DISCUSS	Please share this with family members so they can talk with their healthcare providers and learn more.
Reach Out	RESOURCES	<ul style="list-style-type: none"> • Amyloidosis Foundation amyloidosisresearchfoundation.org • National Society of Genetic Counselors nsgc.org • Canadian Association of Genetic Counsellors cagc-accg.ca • Genetic Information Nondiscrimination Act (GINA) ginahelp.org

Amyloidosis in the Family

Even though your genetic testing was negative, amyloidosis can run in families. All close family members of someone with amyloidosis (like parents, brothers, sisters, children) should talk with their healthcare providers about screening.

Please discuss this information with your healthcare providers. The field of genetics is continuously changing, so updates related to your genetic testing results and/or medical management options may be available over time. This information is not meant to replace a discussion with a healthcare provider, and should not be considered or taken as medical advice.

