

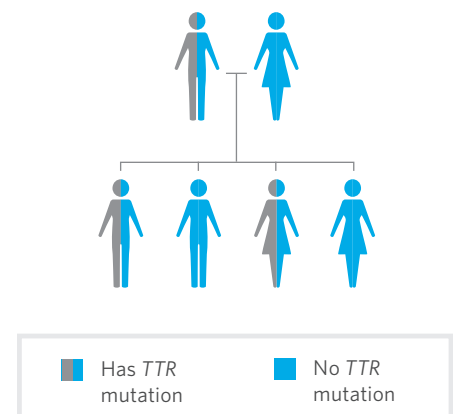
Understanding Your Positive Transthyretin Amyloidosis (TTR) Genetic Test Result

INFORMATION FOR PATIENTS WITH A **PATHOGENIC MUTATION OR VARIANT THAT IS LIKELY PATHOGENIC**

Result	POSITIVE	Your testing shows you have a pathogenic (disease-causing) mutation, or a variant that is likely disease-causing, in a gene that causes transthyretin amyloidosis. Both, mutations and variants that are likely disease-causing, should be treated as the same type of positive result.
Gene	DEFINITION	Everyone has two copies of each gene. We get one copy from each of our parents. Mutations (changes in the gene, like spelling mistakes) in one copy of the <i>TTR</i> gene can cause hereditary transthyretin amyloidosis.
Diagnosis	HEREDITARY TRANSTHYRETIN AMYLOIDOSIS	Hereditary transthyretin amyloidosis is a multisystem disease that most often affects the nervous system, heart, kidneys, and eyes.
Management Options	FOR PATIENTS WITH HEREDITARY TRANSTHYRETIN AMYLOIDOSIS	Treatment options include: medications, surgery, pacemakers, or liver transplantation. Talk to your doctor about which may be right for you.
Family Members	50/50 CHANCE	Your close relatives (like your parents, siblings, children) have a 50/50 chance of inheriting the <i>TTR</i> mutation you carry. Other family members (like your aunts, uncles, cousins) also have a chance of carrying this mutation. Any of your relatives can be tested for the mutation.
Next Steps	DISCUSS	Please share this with family members so they can talk with their doctors 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

TTR mutations in the Family

There is a 50/50 chance to pass on a mutation in *TTR* to your sons and daughters. The image to the right shows that both men and women can carry and pass on these mutations.



Please discuss this information with your healthcare provider. The field of genetics is continuously changing, so updates related to your TTR result, medical recommendations, and/or potential treatments 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.