

# Understanding Your Positive Transthyretin Amyloidosis (TTR) Genetic Test Result

INFORMATION FOR PATIENTS WITH A **PATHOGENIC MUTATION OR VARIANT, LIKELY PATHOGENIC**

Result	<b>POSITIVE</b>	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	<b>DEFINITION</b>	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	<b>HEREDITARY TRANSTHYRETIN AMYLOIDOSIS</b>	Hereditary transthyretin amyloidosis is a multisystem disease that most often affects the nervous system, heart, kidneys, and eyes.
Management Options	<b>FOR PATIENTS WITH HEREDITARY TRANSTHYRETIN AMYLOIDOSIS</b>	Treatment options include: medications, surgery, pacemakers, or liver transplantation. Talk to your doctor about which may be right for you.
Family Members	<b>50/50 CHANCE</b>	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	<b>DISCUSS</b>	Please share this with family members so they can talk with their doctors and learn more.
Reach Out	<b>RESOURCES</b>	Amyloidosis Foundation <a href="http://amyloidosisresearchfoundation.org">amyloidosisresearchfoundation.org</a> National Society of Genetic Counselors <a href="http://nsgc.org">nsgc.org</a> Canadian Association of Genetic Counsellors <a href="http://cagc-accg.ca">cagc-accg.ca</a> Genetic Information Nondiscrimination Act (GINA) <a href="http://ginahelp.org">ginahelp.org</a>

## 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.

