Ambry Genetics®

Understanding Your Positive Transthyretin Amyloidosis (TTR) Genetic Test Result

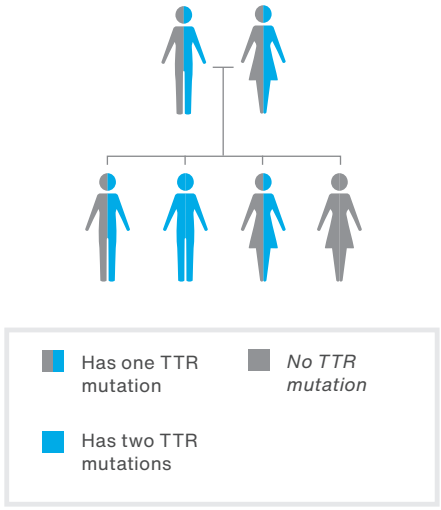
INFORMATION FOR PATIENTS WITH TWO **PATHOGENIC MUTATIONS** OR **VARIANTS, LIKELY PATHOGENIC**

Result	**POSITIVE**	Your testing shows that you have two pathogenic (disease-causing) mutations, or variants that are likely disease-causing, in a gene that causes hereditary transthyretin amyloidosis. Both of these 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 at least one copy of the *TTR* 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 healthcare providers about which may be right for you.
Screening Options	**FAMILY MEMBERS**	Options for screening and early detection for hereditary transthyretin amyloidosis include: physical exams, renal function evaluations, electrocardiograms (EKGs), or imaging studies such as echocardiograms and MRIs. Talk to your healthcare providers about which options may be right for you and/or your family.
Next Steps	**DISCUSS**	Please share this with family members so they can talk with their healthcare providers and learn more.
Reach Out	**RESOURCES**	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

Your close relatives (like your parents, brothers, sisters, children) have at least a 3/4 chance of having at least one of the mutations that you carry, and other family members (like your aunts, uncles, cousins) may also have it. Your relatives can now be tested for these same mutations, if they wish.

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.



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