Ambry Genetics

Understanding Your Positive Genetic Test Result for Thoracic Aortic Aneurysms/Dissections (TAAD) or Related Conditions

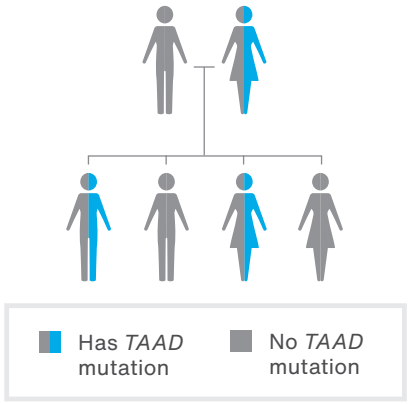
INFORMATION FOR PATIENTS WITH A **PATHOGENIC MUTATION** OR **VARIANT THAT IS LIKELY PATHOGENIC**

Result	POSITIVE	Your (or your family member's) testing shows that you have a pathogenic (disease-causing) mutation or a variant that is likely pathogenic in a gene that causes TAAD or a related condition. 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 one copy of any of the genes in this test can cause TAAD or a related condition.
Diagnosis	SYNDROME	A syndrome is a group of medical signs and symptoms that happen together in one person or family. Syndromes can include TAAD and other health problems. Examples are Marfan syndrome and Loeys-Dietz syndrome.
Diagnosis	FAMILIAL ISOLATED TAAD	Some gene mutations that cause TAAD do not involve a syndrome. This means that they can still run in families, but do not cause other health problems besides TAAD.
Management Options	PATIENTS WITH TAAD OR RELATED CONDITIONS	Treatment options may include: medications, surgery, or avoiding certain athletic activities. Your doctor may also recommend tests for other medical problems that are sometimes associated with your specific test result. Talk to your doctor about which options may be right for you.
Screening Options	FAMILY MEMBERS	Options for screening and early detection include: physical exams, echocardiograms, or cardiac MRI. Talk to your doctor about which options may be right for you and/or your family.
Next Steps	DISCUSS	Share this with family members so they can talk with their doctors and learn more. They can now be tested for this same mutation, if they choose to.
Reach Out	RESOURCES	▪ Ambry's Cardiology Site for Families patients.ambrygen.com/cardiology ▪ National Society of Genetic Counselors nsgc.org ▪ The Marfan Foundation marfan.org ▪ Loeys-Dietz Syndrome Foundation loeysdietz.org ▪ The Ehlers-Danlos National Foundation ednf.org ▪ Genetic Information Nondiscrimination Act (GINA) ginahelp.org
Participate	RESEARCH OPPORTUNITIES	You may wish to contact Dr. Dianna M. Milewicz and her research staff of the University of Texas Health Science Center at Houston, regarding a study of gene changes that can lead to TAAD and related diseases. Participation in research is optional. You may contact the study office at (713) 500-6715 or JRRP.research@uth.tmc.edu. For more information about the John Ritter Foundation, you may visit johnritterfoundation.org.

TAAD and Related Conditions in the Family

Most gene mutations that cause TAAD are passed down in families in an autosomal dominant pattern. This means that your close family members (like your parents, brothers, sisters, children) have a 50/50 chance to also have it. Your relatives can now be tested for this same mutation. Those who DO NOT have this mutation may not be at risk for TAAD and may avoid unneeded screening.

Please talk with your doctor or genetic counselor about this. The field of genetics is continuously changing, so updates related to your 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.



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