

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

INFORMATION FOR PATIENTS

Result	NEGATIVE	Your (or your family member's) testing did not find any disease-causing mutations. 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 one of these genes, you are not likely at increased risk for TAAD or a related condition.
Diagnosis	NO CHANGE	This testing does not change your diagnosis. If you have been diagnosed with TAAD or a related condition, that remains the same.
Further Testing	DISCUSS	More genetic testing may be right for you. Please talk about this with your doctor or genetic counselor.
Management Options	PATIENTS WITH TAAD OR RELATED CONDITIONS	Treatment options may include: medications, surgery, or avoiding athletic activities. Your doctor may also recommend tests for other medical problems that are sometimes associated with TAAD. 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, if any, options may be right for you.
Next Steps	DISCUSS	Share this with family members so they can talk with their doctors and learn more.
Reach Out	RESOURCES	<ul style="list-style-type: none"> • 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 loeydietz.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

Even though your (or your family member's) genetic testing was negative, TAAD could still be running in your family. All close family members of someone with TAAD (like parents, brothers, sisters, children) should talk with their doctor about 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.

