What is the John Ritter Research Program?

The John Ritter Research Program (JRRP) is a collaboration between The University of Texas Health Science Center at Houston (UTHealth) and the John Ritter Foundation for Aortic Health to identify the genetic causes of thoracic aortic aneurysms and dissections (TAAD) and other vascular diseases. Our mission is to prevent premature deaths due to aortic dissections by identifying genetic risk factors and individuals at risk for aortic dissections. Additionally, we seek to improve treatment for thoracic aortic disease and to increase public awareness about genetic and other risk factors for acute aortic dissections.

Causes of Thoracic Aortic Aneurysm and Aortic Dissection

Two established risk factors for thoracic aortic aneurysms and dissections are poorly controlled high blood pressure and the presence of a bicuspid aortic valve. In recent years, a genetic predisposition to TAAD has emerged as a major factor causing an increased risk for thoracic aortic aneurysms and dissections.

Familial Thoracic Aortic Aneurysm and/or Dissection (FTAAD)

TAAD can “run in families” and the term familial TAAD or FTAAD is used whenever two or more family members are affected with thoracic aortic aneurysm or dissection. Often these individuals do not have features of a genetic syndrome. One in five individuals with TAAD have a close relative with TAAD. Therefore, having a family history of thoracic aortic aneurysm or aortic dissection is a risk factor for TAAD. In families with FTAAD:

• Both men and women can be affected.
• TAAD is passed down from one generation to the next in a pattern known as autosomal dominant inheritance. This
means each child of an individual with TAAD has up to a 50% chance of inheriting the risk to develop TAAD.

- Having a genetic predisposition for TAAD does not mean that one is certainly going to develop aortic disease, but rather one has an increased risk to develop an aortic problem and therefore needs to be closely monitored to detect signs of aortic disease.
- Familial TAAD has variable presentation, which means individuals within the same family may have different ages of onset and disease severity.
- TAAD may occur alone or may be associated with other blood vessel and heart problems, for example a bicuspid aortic valve or other arterial aneurysms.

Genetic Syndromes

Some genetic syndromes known to be associated with TAAD are Marfan syndrome, Loeys-Dietz syndrome, the vascular form of Ehlers-Danlos syndrome, and Turner syndrome.

Environmental Factors

Uncontrolled high blood pressure, smoking, weight lifting, trauma to the aorta, and certain inflammatory diseases are some of the factors that can increase the risk for an aortic aneurysm or dissection. These factors may act alone or interact with genetic factors to cause an aortic aneurysm or dissection.

Our Research Studies

Dianna M. Milewicz, M.D., Ph.D., Director of the John Ritter Research Program in Aortic and Vascular Diseases at UTHealth, is an internationally recognized expert in the genetics of aortic and vascular diseases. She works with a team of clinicians, scientists, and genetic counselors on a number of research studies that are focused on learning more about the genes that cause aortic aneurysms and dissections and other vascular diseases. Our studies have already identified genes for FTAAD, and the genes are being used by doctors to identify individuals at increased risk for aortic disease.

If you have been diagnosed with a thoracic aortic aneurysm or dissection, please contact us to take part in our research study. We can be reached at (713) 500-7072 or info@JohnRitterResearchProgram.org. One of our genetic counselors will explain the study, determine whether you qualify to participate in the study, and help you enroll if you decide to participate.
What Will I Have to Do to Participate in the Study?

Enrollment in research will take approximately 30-60 minutes of your time. Note that you can take part in our research studies without leaving your house. You will be asked to provide the following in order to take part in research.

1. **A DNA sample.** You will be asked to provide a saliva sample that will be used to isolate your DNA for genetic studies. A saliva self-collection kit and the research consent forms will be mailed to your home. The genetic counselor will arrange a time to explain the study to you via telephone and give you an opportunity to ask questions. If you decide to participate, you will be asked to complete the consent forms, collect a sample, and mail these back to our laboratory. In some cases, you may be asked to provide a blood sample (approximately 2 tablespoons) by undergoing a blood draw at your doctor’s office.

2. **Medical Records.** You will also be asked to provide health information related to the research study. This information can be obtained from your medical records and you may be asked to provide authorization to release this information for research.

What Are the Benefits?

Our research may not help you or your family members directly, but may shed some light on the causes of aneurysms and dissections that affect you or your family. You should not expect to personally benefit from this research. The main reason you may want to participate is to help our researchers understand the causes of aortic problems so that we can find better ways to prevent, detect, and properly treat thoracic aortic disease to prevent premature deaths from aortic dissections.

How Can I Help?

- **Contact us** to find out if you or your family qualifies to participate in our study. We can be reached at (713) 500-7072 or info@JohnRitterResearchProgram.org.
- **If you are a physician or other healthcare provider** who has a patient with a personal or family history of TAAD, please provide our contact information to the patient or contact Dr. Milewicz or one of our genetic counselors about referring to our study.
- **Become an aortic disease advocate** by helping us raise awareness and educate others about familial thoracic aortic disease.
- **Donate to support the John Ritter Foundation and John Ritter Research Program** and help us prevent further premature deaths due to acute aortic dissection. 
  (www.JohnRitterFoundation.org/donate-today/ or
  www.JohnRitterResearchProgram.org/donate/).

For questions or to speak with a genetic counselor, email info@JohnRitterResearchProgram.org

or call

(713) 500-7072

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**UTHealth**

The University of Texas Health Science Center at Houston

**Medical School**

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**John Ritter Research Program in Aortic & Vascular Diseases**