January 2020

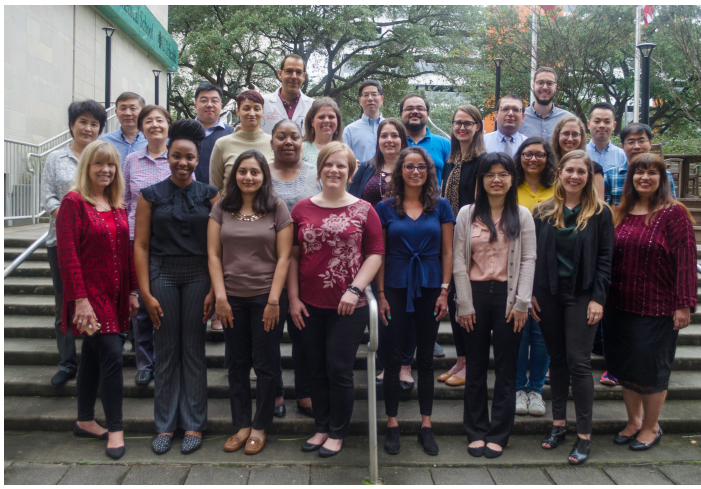
Newsletter

JOHN RITTER RESEARCH PROGRAM IN AORTIC & VASCULAR DISEASE

THANK YOU FOR YOUR SUPPORT IN 2019!

HAPPY NEW YEAR FROM THE JRRP!

We wish you all a happy and healthy start to the new decade and thank you for your continued support for the John Ritter Research Program. As we work to understand the relationships between genetics and aortic and vascular disease, we would like to take the opportunity to use this newsletter as a platform to provide quarterly updates on the efforts our team and this community have made toward improving outcomes for patients and families.



AORTIC DISSECTION AWARENESS DAY SEPTEMBER 19

This past year, Dr. Milewicz traveled to the UK to speak on the critical need for identification and early diagnosis of aortic dissection to save lives. The THINK AORTA campaign is dedicated to raising awareness among healthcare professionals.

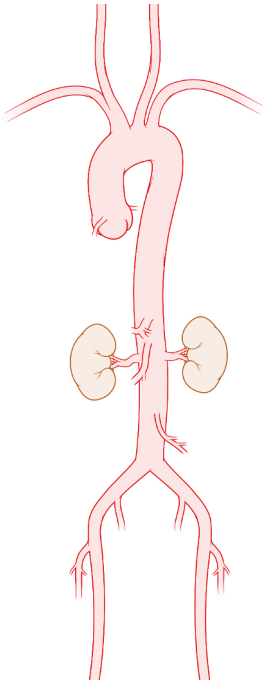
2019 was a busy year for the John Ritter Research Program. With the help of our collaborators, patients, and research participants, we made major strides in advancing aortic disease research. Members of our lab published several key research articles in 2019 including a review of the 'Genetics of Thoracic and Abdominal Aortic Diseases' in the well known *Circulation Research* journal. With this momentum, we plan to accomplish even more in 2020!

Recent research findings published in the *Journal of the American College of Cardiology* shows the role genetics contributes to aortic dissections in patients with no family history of aortic disease. The study which included a review of 355 patients by Guo et al., highlights that almost 10% of patients with a history of "sporadic" aortic dissection have an identifiable genetic cause. Sporadic dissection refers to patients diagnosed with an aortic dissection who don't have a family history or any physical characteristics we see in patients with Marfan or Loeys-Dietz syndrome, such as long fingers or scoliosis.

While it is well-known that up to 20% or 1 in 5 patients diagnosed with an aortic aneurysm or dissection have a family member who is also affected, data from this study shows that people without a family history still need

genetic consultation and testing. "It's a common misconception that if you don't have a family history of aortic disease or features of Marfan syndrome, the disease is not genetic," said JRRP Genetic Counselor, Alana Cecchi.

The THINK AORTA campaign, initially launched through patient and provider advocacy efforts in the UK and Ireland, will be expanding to the US in 2020 under the guidance of Dr. Dianna Milewicz and expert collaborators.



The goal of the campaign is simple, yet extremely important. Aortic dissections often lead to fatalities if not diagnosed in a timely manner by medical professionals. THINK AORTA is focused on cultivating awareness among emergency medical providers to improve the outcomes for patients with aortic dissections. THINK AORTA is scheduled to launch across US hospitals this year with the help of patient and provider advocates. To make this successful, we need advocates to work with their local providers and hospitals to raise awareness and promote the THINK AORTA campaign materials in emergency departments across the country. If you are interested in participating in the campaign, you can contact info@johnritterreasearchprogram.org.

THINK AORTA

AORTIC IMAGING FOR FAMILY MEMBERS

Guidelines from the American College of Cardiology Foundation and American Heart Association recommend thoracic aortic imaging for first-degree relatives of patients with thoracic aortic aneurysms or dissections to identify people with asymptomatic disease. This may include imaging such as an echocardiogram or chest CT scan. Talk to your healthcare provider about how you can get screened.

PREVENTATIVE SURGERY FOR GENETICALLY TRIGGERED THORACIC AORTIC DISEASE

PATIENT STORY SPOTLIGHT

After Zowie Claudio lost her father at age 35 and brothers at ages 23 and 18, she and her mother knew they needed to take action.

A member of the Claudio family contacted Dr. Dianna Milewicz, Director of the John Ritter Research Program at UTHealth in 2005 after the death of Zowie's father. Several members of the family enrolled in Dr. Milewicz's research study focused on identifying genes that predispose to aortic aneurysm and dissection.

"WE NEEDED TO DO SOMETHING."

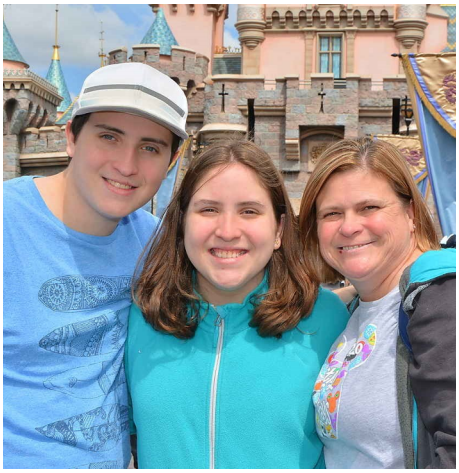
At the time when they entered the study, the underlying genetic cause for the family's aortic disease was unknown. Several years later, through participation in the study, the family's causative gene mutation was identified, an alteration in the *PRKG1* gene. Researchers in Dr. Milewicz's Lab determined that Zowie, her father, and two brothers, all carried a *PRKG1* gene mutation, responsible for the aortic dissections in their family.

Zowie and her mother Angela, enjoying a trip to Alaska after recent aortic surgery.

"Being able to pinpoint the genetic cause of disease is very powerful. It allows healthcare providers to use a gene-based medical management strategy -- the goal of personalized medicine," Milewicz said. "Knowing the gene enables us to test family members who are at-risk to prevent additional deaths from dissections."

Armed with the information that Zowie, 16, carries the gene mutation that predisposed to the deadly aortic dissections her father and brothers suffered, the family traveled to Houston, Texas to undergo a preventative aortic repair in the hands of Dr. Anthony Estrera, Chair of Cardiothoracic and Vascular Surgery at UTHealth. "We needed to do something. We couldn't just hope that everything would be all right," Zowie's mother, Angela, said.

Zowie's surgery was a success, reducing her risk of death from aortic dissection. She's back at school, enjoying time with friends and family.



Zowie with her mom and brother, at Disney World.

HOW TO GET INVOLVED IN RESEARCH

CONTACT US TO LEARN HOW YOU CAN PARTICIPATE IN ONE OF OUR RESEARCH STUDIES

Email us at **JRRP.research@uth.tmc.edu.** You may be eligible to participate in research if:

1. You have been diagnosed with a thoracic aortic aneurysm or dissection *and* have a family member who has had an aneurysm or dissection.
2. You have been diagnosed with a thoracic aortic dissection at age 55 years or younger.
3. You are a woman who had a dissection during childbearing years (under 50) or during/after pregnancy.

RESOURCES

Participating in genetic research is not a replacement for consultation with a genetics professional or clinical genetic testing. You can find an expert in your area through the Marfan Foundation, National Society of Genetic Counselors, and the American Board of Genetic Counseling.

FOLLOW US ON SOCIAL MEDIA

Please consider supporting our research to propel this mission forward.

Your gift makes it possible for our team of dedicated researchers to investigate the genetic causes of aortic aneurysms and dissections to improve outcomes for patients and families.

CALENDAR

MARCH 17

Aortic Hope hosts "Ask a Dr." with Dr. Prakash, UTHealth Cardiologist specializing in thoracic aortic disease.

APRIL 4

The John Ritter Foundation and Aortic Hope will host a joint symposium in Baltimore for the patient community. Watch for more event details in coming months.

JULY 9–12

The 36th Annual Marfan Foundation Conference will be held in Boston, MA.

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