Raising Aortic Disease Awareness


On November 2, 2014, Frank R. Arko III, MD, will be running the TCS New York City Marathon on behalf of the John Ritter Foundation for Aortic Health. *Endovascular Today* spoke with Dr. Arko and Dianna M. Milewicz, MD, PhD, Director of the Ritter Foundation, about raising awareness for aortic disease and the future of aortic research.

What are the goals of the John Ritter Foundation for Aortic Health?

Dr. Milewicz: In 2004, Amy Yasbeck contacted me about John Ritter's death due to aortic dissection, and she was already in the process of forming the John Ritter Foundation. We have worked together to build on the mission of the John Ritter Foundation. Amy is particularly passionate about making sure that individuals who have survived an acute aortic dissection or lost a loved one to dissection obtain information and proper genetic counseling about the risk of aortic disease in other family members.

The Ritter Foundation mission statement is three-pronged. The first is to increase awareness, both in the general public and in the doctors caring for patients with the disease. When the treatment guidelines were published, the American Heart Association, American College of Cardiology, cardiovascular surgeons, radiologists, and emergency department doctors all approved these treatment guidelines. We then assembled the Thoracic Aortic Disease Coalition to increase awareness and that involved the Ritter Foundation, the Marfan Foundation, the Bicuspid Aortic Valve Foundation, and various other foundations that share concern for an increased risk for dissection. We created the Ritter Rules, which are a set of reminders to recognize, treat, and prevent aortic dissection, and we are in the process of putting the treatment guidelines on a website, so these recommendations are completely searchable for physicians and laypeople. If a doctor sees somebody with a 4.5-cm ascending aneurysm, the answers they are looking for are on our site. Like patients, doctors are Googling for information on diseases, so we want to be the go-to website for aortic disease information. Most of the information is distilled and updated from the treatment guidelines.

Our second goal is to provide support to patients and families who have either lost a loved one or survived a dissection, to make sure that they get the proper grief and genetic counseling. We want them to be aware that there may be other family members at risk. We run the counseling for the John Ritter Foundation out of my UTHealth office, and it is handled by one of my genetic counselors.

Third, our research goal is to make sure we identify genes and other factors that increase the risk of a dissection. We seek to better understand the link between these genetic predispositions and environmental factors (eg, hypertension and weight lifting, respectively) that may lead to an acute dissection. We can then develop better drugs and ways to identify those at risk as we gather this information.

If a genetic predisposition isn't known, then we can offer imaging to the family members and recommend echo or magnetic resonance imaging (MRI). The echo can visualize the very first part or aortic root very well, but it doesn't show the ascending aorta particularly well. Patients then undergo MRI or CT, but we push for MRI due to the radiation associated with CT.

Dr. Arko: The Ritter Foundation has a mission to increase awareness and educate about aortic disease. In our own center, we evaluate and treat patients with bicuspid aortic valve disease, ascending aortic aneurysms, coarctation, and type A and B aortic dissections utilizing a multidisciplinary approach. This multidisciplinary approach allows for awareness to be increased throughout the hospital system, emergency departments, and primary care physicians. This education allows for earlier diagnoses and appropriate intervention when indicated. When treated electively, these patients actually do quite well, with a relatively low rate of morbidity or mortality compared to when they are operated on emergently.

What protocols should be in place to discern this disease from other conditions, particularly in the emergency department setting?

Dr. Arko: As a vascular surgeon, it's an easier diagnosis for me to make because it's something I manage daily.

But for emergency department physicians who see several thousand patients on a yearly basis, it can be a difficult diagnosis to make. Some may not even understand what a dissection is and its risk of morbidity and mortality when treated inappropriately. It can present in a variety of different ways, and unless your suspicion is high, it can be missed. One of my goals, as well as that of the Ritter Foundation, is to increase awareness for these acute aortic syndromes. If you diagnose aortic dissection early, you save lives, and if you miss the diagnosis, patients are at serious risk of death.

In patients with aortic aneurysms (either in the thoracic or abdominal component) and/or dissections, I think it's very important to follow these patients with lifelong surveillance, including the aortic valve.

Dr. Arko with his wife and youngest son earlier this year.

Dr. Milewicz: Proper identification of patients with acute aortic dissections in the emergency department is an issue that the John Ritter Foundation and the Thoracic Aortic Disease Coalition are working on. The American Heart Association and American College of Cardiology guidelines for the diagnosis and management of patients with thoracic aortic disease included high-risk clinical features to assist in the early detection of acute aortic dissection out of all the people who walk into the emergency department with chest pain. These clinical markers included asking the patient about a history of a bicuspid aortic valve, Marfan or related syndromes, or a family history of aortic aneurysms or aortic dissection. Another marker of dissection is the features of the pain, because dissection pain (sharp, tearing, migrating pain) is very different from a heart attack or other type of pain. During the physical exam, patients with an acute dissection may have pulse deficits and other features of dissection. These findings can determine who needs to go to the next step and undergo aortic imaging for an acute aortic dissection.

We're trying to get these clinical risk markers "hardwired" into emergency departments by working through the people who work with the electronic medical records and the risk-management offices at hospitals. There is not a doctor out there who wants to miss a dissection, but we need to get the right questions hardwired into the screening process for people who come in with chest pain. Ideally, we would have a biomarker that worked like the biomarkers for acute myocardial infarction, which is an area of current research.

What other screening protocols do we need to help identify asymptomatic patients?

Dr. Milewicz: Somebody who presents with aortic disease at a young age or with features of Marfan syndrome with a family history of aortic disease should undergo genetic testing. Our work, along with the work of other investigators, has identified 10 genes that lead to an inherited predisposition for the disease. All these genes can be sequenced at once. If we find a genetic variant in the family causing disease, it becomes very powerful information. We can then go through the family to see who is at risk for the disease and set up an imaging protocol where they are imaged routinely for an aneurysm. If an aneurysm is identified, medication is started, and patients undergo yearly imaging. If it becomes necessary, these patients can be sent to aortic surgical repair to prevent a dissection. Genetic screening also tells us whether people are at risk for problems with other arteries, such as aneurysms in the arteries in the head; some of the genes cause a risk for not only aortic disease, but also other vascular diseases.

Genetic testing costs have fallen dramatically, and we can sequence all of the known genes for less than an MRI or CT. This is why we start with genetic testing first. Echo, MRI, and CT costs vary based on where you go. Echo is useful if you know if the aneurysm involves the aortic root at the level of the sinuses of Valsalva. Echo is a good way to follow those aneurysms because there is no radiation, and usually, it is less expensive than MRI and CT.

Although genetic testing is usually covered by insurance, we do have more trouble getting the genetic tests covered than the aortic imaging. There are families with hundreds of people in them at risk, so if we can find the gene, we can lower the amount of necessary imaging. Once again, if we know the gene, we also know whether routine echo imaging is enough or if we have to do MRI or CT imaging, as well as the frequency of imaging. Perhaps most importantly, the gene will also indicate the timing of surgical repair to prevent aortic dissections.

In people with Marfan syndrome, we can watch the aorta grow to a diameter of 5 cm, and the risk for dissection is very low, especially if they're not lifting weights, their blood pressure is controlled, and they are on beta-blockers or other medication. However, in someone who has other mutated genes, we know we have to be much more aggressive about the surgical repair, because there can be a very high risk for dissection, even when the aneurysm is < 5 cm.

What can hospitals and practices do to raise awareness about aortic disease?

Dr. Arko: Hospitals and practices can do several things. First, they can educate their primary care physicians as to what the risk factors are for developing aortic disease, both in the thoracic and the abdominal area. Second, it's important to get larger centers to develop an aortic center that has a multitude of specialists involved in the care of these patients. Furthermore, it is also important to improve the education of those who are or may be at risk with a strong family history. Family members with a strong history of aneurysms/dissections and congenital heart disease, including bicuspid aortic valves and coarctations, should undergo appropriate imaging for screening. Lives can be saved with appropriate medical therapy, surveillance, and early intervention when indicated.

As technology continues to improve, hopefully, we will have better ways to treat these patients from a less invasive standpoint.

Dr. Milewicz: Patients with an inherited predisposition for thoracic aortic disease should undergo genetic counseling and testing. The John Ritter Foundation can help provide information on which family members need to be screened and offer a referral to a geneticist to follow up with the family and genetic testing when appropriate.

Hospitals should focus on hardwiring the emergency department system so that they don't miss an acute dissection. It is difficult to disseminate this information because so many other organizations are trying to make hospitals aware of their disease, but I think aortic disease in particular is a situation in which a concerted effort to make sure that these screening protocols are set up in the emergency department would lead to acute aortic dissection being diagnosed in a timely manner.

What are some examples you have encountered that illustrate why we need to raise awareness?

Dr. Arko: I have treated a number of patients with a familial history of thoracic aortic aneurysmal disease, as well as dissections. I suggest to my patients that their relatives get screened with appropriate imaging studies, which may include echocardiography, CTA, or MRI.

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It's also important to talk to patients about risk factor modifications from a cardiovascular disease standpoint, whether that be peripheral arterial disease, aneurysmal disease, and/or a combination of the two. Certainly, cessation of smoking is an extremely important component in any discussion with patients at risk for cardiovascular disease. If there is a strong family history of aneurysmal disease, genetic testing is important to evaluate for potential connective tissue disorders.

How did the Ritter Foundation decide to form a marathon team as a means to raise awareness? What will the raised money fund?

Dr. Milewicz: We approached the organizers of the New York Marathon for the John Ritter Foundation to be a designated charity for the marathon. This is the third year that we have had a team in the New York Marathon, and we have had an increasing number of slots for our team each year. The team members all have a connection to thoracic aortic disease and have done an outstanding job raising money. The money raised goes to further the mission of the John Ritter Foundation—dedicated to saving the lives of people at risk for acute aortic dissection through increased awareness, education, and research.

What is your anticipated finish time?

Dr. Arko: My current personal record is 3:43 in the Marine Corps Marathon in Washington, DC. I am attempting to go under 3:30 for this race. Certainly, this race is crowded, and weather always plays a factor in one's time. ■

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