Hereditary Hemorrhagic Telangiectasia

Jesse Jones MD and Theresa Caridi MD discuss hereditary hemorrhagic telangiectasia. In this physician round table, they explore diagnosis and treatment of this genetic disease that can result in nosebleeds, breathing difficulties, and stroke.

Featured Speaker:

Jesse Jones, MD | Theresa Caridi, MD

Jesse Jones, MD Specialties include Diagnostic Radiology, Endovascular Neurosurgery, Interventional Neuroradiology, Neuroradiology and Neurosurgery.

Learn more about Jesse Jones, MD
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Theresa Caridi, MD, FSIR, is an Associate Professor and the Division Director of Vascular and Interventional Radiology at the University of Alabama at Birmingham (UAB). After attending the University of Florida for medical school and radiology residency, Dr. Caridi completed a fellowship in Vascular and Interventional Radiology at the University of Pennsylvania. The first seven years of her career were spent at Georgetown in Washington, D.C., before joining the faculty at UAB.

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Melanie Cole (Host): Welcome to UAB Med Cast. I'm Melanie Cole. And today we're discussing hereditary hemorrhagic telangiectasia. Joining me in this panel is Dr. Theresa Caridi. She's the Director in the Division of Interventional Radiology, Vice Chair of Interventional Affairs in the Department of Radiology and an Associate Professor at UAB Medicine and Dr. Jesse Jones. He's an Assistant Professor and an Interventional Neuroradiologist at UAB Medicine. Doctors, thank you so much for joining us today. Dr. Caridi, I'd like to start with you. Tell us a little bit about hereditary hemorrhagic telangiectasia or HHT. Are there different types? Tell us a little bit about this condition.

Theresa Caridi, MD (Guest): So, HHT or hereditary hemorrhagic telangiectasia is an autosomal dominant disorder. So, it's a genetic disorder. It occurs in about one in five to 8,000 individuals. And the key characteristics of it are that it results in arterial venous malformations and telangiectasias at various sites. There are various forms of it, but all of the genes that are known to be affected, involve a signaling cascade of molecules that are crucial to vascular

development, which is why these individuals end up with arterial venous malformations, which we can just refer to as AVMs and also telangiectasias in places all over the body. So, at one characteristic spot, for instance, is in the lungs and that's known as a pulmonary arterial venous malformation, but these malformations can occur in several areas of the body and need various types of treatment depending on where they occur.

Jesse Jones, MD (Guest): Another common manifestation of HHT is nose bleeding. Many people typically present with nose bleeding, typically around a time of older childhood or adolescence and the nose bleeds can be quite severe, requiring treatment, ER visits and even surgical procedures to address their nose bleeding. Because HHT can affect so many body sites, including the lungs, the heart, the GI tract, and even the brain and spine, it's important that these people are assessed in a center well-versed in HHT with multidisciplinary cooperation.

Host: Well, thank you both for telling us about that. So, Dr. Jones, I'd like you to expand a little bit on the affected populations and more of those symptoms. You talked about nosebleeds, but when does this seem to present itself for referring physicians and internists and providers, what would they notice and when.

Dr. Jones: Well, because like Dr. Caridi mentioned, the disease is actually quite common. You know, up to one in 5,000 people tend to be affected, but nosebleeds are much more common than that. And so most people with nosebleeds, don't just assume they have HHT and the doctors don't assume they have it either. So, it does take a knowledge of the disease in order to get it kind of in a physician's mind that, hey, this patient with recurrent nose bleeding may have HHT, and they should possibly be worked up for it.

There's a series of criteria that we utilize to diagnose these people, including history and physical and genetic testing, now, which is very helpful, but like I mentioned previously, the nosebleeds tend to start fairly early in life and because it's genetic, there's typically a family history of nosebleeds as well, which is very important in the history.

Host: That is important. And what a great point that you made. So, Dr. Caridi, what is involved in diagnosis? Obviously a good history is important, but tell us a little bit about why it's sometimes difficult to diagnose.

Dr. Caridi: Yeah, sure. So, the difficulty in diagnosing HHT is just that it manifests in different individuals and various ways. So, while some patients, most typically one of the manifestations is nosebleeds or epistaxis, not all patients have that. And so they may have a different variant and a different manifestation. As we said before, it can affect many regions of the body. We do use the Curacao criteria, which are four criteria that come along with the patient's history, that can really lend towards the diagnosis, if these are positive or if a few of them are positive. Those include multiple telangiectasias of the skin and mucous membranes, repeat episodes of spontaneous nosebleeds, or epistaxis, visceral vascular malformations and a family history in a first degree relative. So, those are the four Curacao criteria. If patients manifest one or more of these, they should be considered for this disease process. Sometimes this may be an incidental finding on an imaging study, such as a pulmonary AVM. And that would meet one of these Curacao criteria.

So, patients who present with epistaxis, nose bleeding, or they have a manifestation of one of these arterial venous malformations on imaging, an unusual age or presentation for a stroke or other neurologic event, like a TIA, or a family history of any of these things, those patients should be considered for HHT.

Host: Dr. Jones, tell us about some of the standard therapies you would use once you've discovered someone has HHT.

Dr. Jones: Yeah, the therapies are quite tailored to the presentation and as Dr. Caridi mentioned, the presentation can be quite protean. We typically address people symptomatically and also through a comprehensive screening program. So, patients who present with repeated bothersome nose bleeds, will typically see one of our ENT

surgeons here at UAB, where we focus mainly on preventative strategies and trying to avoid and more invasive surgical procedures.

These would include things like keeping the patient's mucus membranes moist and well-hydrated, there's things like inhaled, Avastin, which can also be very helpful to prevent recurrent nose bleeds. And if patients do end up needing a surgery, the surgery is typically as minimally invasive as possible. It would be an endoscopic approach using what's called a COBLATOR to really close off some of these telangiatasias in the nose without damaging the nasal septum or other delicate areas of the nasal mucosal membranes. Dr. Caridi is an expert in pulmonary AVMs and she would treat these people through an endovascular technique. I can let her discuss more fully, but as part of the comprehensive screening program, our patients would typically also see a pulmonologist. They would see a cardiologist, a GI doctor for other visceral AVMs and screening for those including capsule endoscopy, as well as a geneticist and a hematologist to manage anemia.

Host: Dr. Caridi, would you like to expand on what Dr. Jones just said?

Dr. Caridi: Yeah, sure. I'll expand on the pulmonary arterial venous malformation aspect of things. If you see cana patient with an incidental pulmonary AVM on imaging, or a symptomatic one, about 65% or even more of those patients actually have HHT so it's an unusual condition to find a pulmonary AVM that's not associated with HHT actually, although it does happen, there are some that are not at all associated with HHT. The point there being, if a physician or a provider has a patient who undergoes CT imaging for some reason, and an incidental pulmonary AVM is found, it should trigger that provider, hopefully with the knowledge provided in this podcast to go ahead and refer that patient to be evaluated for HHT.

The good news is that Dr. Jones and I run a simultaneous clinic essentially on the same day, a couple of floors apart, so that patients come whether as an individual or with their entire family to be screened and worked up for HHT, since this is a genetic disorder, sometimes these patients will prefer to arrive with their families, as a unit so that all of the family can be screened and appropriately managed if they have HHT.

Host: Tell us a little bit, Dr. Caridi about that multidisciplinary approach. We've touched on it a few times. How do you all work together? You just explained how you and Dr. Jones do. And that's great information for other providers about bringing in the family for genetic testing and screening and how you're just a few floors apart. Tell us about some of the other providers that are involved in how you all work together for this type of approach.

Dr. Caridi: So, as I mentioned before, Dr. Jones, and I actually see patients on the same day, in the same general area of our clinic. So, we can tend to see patients one right after the other. We manage most of the typical manifestations of HHT, but patients definitely often have to go on to other services as well. Sometimes those are able to be scheduled in that same timeframe within either that day or within a 24-hour period. So, if they're coming from a distance, they can get everything accomplished in one visit. The other part I'd like to mention about this is we also see pediatric patients, which can be afflicted by this disease as well.

And so we can follow them from pediatric population all the way through adulthood. And we can either do that at UAB or at the adjacent children's hospital, depending on what we're doing with each patient and what their presentation is. But all of these things can be, our coordinators work together, both at UAB and at the children's hospital to accomplish this for our patients and their families.

Dr. Jones: Yeah, I think coordination of care is very important in this population. Because there are so many organ systems affected. So, we work closely with a small but dedicated team of specialists to get these patients seen on the same day, or at least than 24 hour period to get a comprehensive evaluation and management plan put together in one session. As bad as COVID has been in the last year, it has some positive effects in terms of being a game

changer in terms of remote access. Before COVID, there was emedicine, but it's really kind of gone to lightspeed in the last several months here. And we've utilized that to our advantage with HHT population to get these people seen sooner, in terms of emedicine visits, which can be a video visit or a phone visit to get some important history and physical exam, and also to coordinate their future care.

Host: Such an interesting topic we're discussing here today. I'd like to give you each a chance for a final thought. Dr. Caridi, what are some investigational therapies you're most excited about?

Dr. Caridi: I'll speak a little bit about pulmonary AVM embolization. I mentioned it a little bit before, but I think there are rapid expansion of devices to be used for the pulmonary AVM embolization. Mostly these involve coils and plugs, but the technology is rapidly advancing. And so we can accomplish the procedures faster, with less radiation and with less sessions for the patient to come back for if they have multiple pulmonary AVMs, for instance. So, I think, in my particular space, I'm excited about the technology and the advancements that are available to us.

Dr. Jones: Yeah, I think medicine is changing fast with HHT in particular. We partnered with the foundation, known as Cure HHT and Cure HHT is a, is an umbrella group that support centers of excellence around the country, which we are currently applying to become such a center to better serve this population. But they also coordinated a lot of important clinical trials. And so at UAB, we'll be starting a trial in conjunction with Cure HHT to look at an investigational agent called politimide is a drug that is taken orally, which has been shown in early studies to significantly reduce the amount of bleeding, particularly from epistaxis, but also bleeding from the GI tract. And we're hopeful by conducting this trial and bringing this drug to HHT sufferers in the Alabama community and throughout the Southeast region that we can significantly improve their quality of life.

Host: What great information and Dr. Jones, do you have a final thought on referring providers and what you'd like them to know about early referral and communication with the referring physician?

Dr. Jones: Well, I think it's very important to have a person to reach and a phone number to call. Dr. Caridi and I are both always available to see new patients and we'd be happy to take any referrals, in the region and take it from there in terms of coordinating their care and working them up.

Host: Thank you both so much for joining us today. And a physician can refer a patient to UAB Medicine by calling the mist line at 1-800-UAB-MIST or visiting our website at uabmedicine.org/physician. That concludes this episode of UAB Med Cast. Please remember to subscribe, rate and review this podcast and all the other UAB Medicine podcasts. I'm Melanie Cole.