Vlad Zaha, MD: 00:18

Welcome to the Hypertrophic Cardiomyopathy Podcast, titled Diverse Spectrum to Presence, Severity and Type of Cardiovascular Symptoms in Patients with Hypertrophic Cardiomyopathy. This is one of a series of podcasts from the American Heart Association, Hypertrophic Cardiomyopathy Initiative, sponsored by Bristol Myers Squibb.

I'm Dr. Vlad Zaha at UT Southwestern Medical Center, where I'm a specialist in multimodality imaging and medical director of the cardio-oncology program. And I'm joined today by Dr. Martin Maron, director of the Hypertrophic Cardiomyopathy Center and Research Institute at Tufts Medical Center, and director of the Chanin T. Mast HCM Center at Morristown Medical Center, New Jersey, and by Dr. Albree Tower-Rader, instructor of medicine at Harvard Medical School and the Hypertrophic Cardiomyopathy Center at Massachusetts General Hospital.

We also have with us a patients' representative Shawn, today, and we are really excited to be able to start a discussion here about the diverse spectrum of symptoms and presentations that the patients can learn about their hypertrophic cardiomyopathy from. So, I would like to invite Dr. Maron to open the discussion and to tell us a little bit about perspective on the patient presentation.

Martin Maron, M...: 01:49

Let's introduce Shawn. Hi, Shawn.

Shawn Lewis: 01:51

Hi.

Martin Maron, M...: 01:51

Thanks for taking the time to do this and to share your story and insights, appreciate it very much. Maybe if it's okay, we could start with you, and you could just begin by telling us a little bit about your story.

Shawn Lewis: 02:05

Okay. I guess about 12 years ago, I started getting sick, a lot of misdiagnosis and I've had pneumonia, bronchitis, asthma, different breathing type of illnesses. And it just continued to get worse, with no relief. And this particular time when I was diagnosed, it had been going on for three months, I had been going to work, can't breathe, take two steps, you can't catch your breath, you can't move, you can't... You're just full off what I've learned later on to be liquid. I went to emergency room after about three months of going back and forth to different doctors, stayed there about 10 hours and then they told me I had cardiomyopathy and heart failure. Stayed in the hospital about three days, getting rid of 22 pounds worth of fluid and

		then that's when I found out, hey, you have these things and you need to start seeing a cardiologist to treat you.
Martin Maron, M...:	03:10	Right. So, maybe we could stop there for just a second because I want to focus for a minute on what you said in the beginning, which is that you had a lot of symptoms and you were not diagnosed with cardiomyopathy for a while with those symptoms.
Shawn Lewis:	03:25	Right.
Martin Maron, M...:	03:26	And I think that raises a really important first principle point here is that, HCM can present with symptoms that overlap with many other cardiovascular diseases, and also non-cardiac diseases, lung problems, as you mentioned, as well. And so, one of the efforts to improve diagnosis is I think, to help improve the education of what symptoms the patients with HCM could present with and so that physicians have a better sense of looking for HCM sooner, rather than having to delay. In this case, it sounds like quite a bit of a delay in diagnosis, which obviously I think we all agree, we're trying to avoid and improve. And so, maybe it's worth just sort of talking a second about those symptoms. It sounds like you had a lot of shortness of breath as part of your presentation, is that correct?
Shawn Lewis:	04:21	Correct.
Martin Maron, M...:	04:21	And that was misdiagnosed essentially, as lung disease, as asthma, is that right?
Shawn Lewis:	04:21	Right.
Martin Maron, M...:	04:27	Yeah.
Shawn Lewis:	04:28	Yes.
Martin Maron, M...:	04:28	So, I think part of the effort here in this HCM educational initiative here is to make the practicing community more aware that shortness of breath can be related to a number of different things, including hypertrophic cardiomyopathy. And then if the patient's not responding to their symptoms initially with [inaudible 00:04:48] therapy, that we need to broaden out the potential differential diagnosis to include heart diseases like HCM. Okay. And the important point there is that HCM prevalence is equal among gender, men and women, it's the

same prevalence. And so, we have to also consider that as well, is that irrespective of whether a patient's presenting with symptoms as a male or female, if they've got shortness of breath or chest discomfort or exertional fatigue or lightheadedness, and we don't have an explanation for another disease, HCM has got to be up there on the top of the list, as well, so that we can help improve the time to appropriate diagnosis and treatment in patients like you, so that you didn't have to suffer probably longer than you had to, which it sounds like was quite a bit of time.

Shawn Lewis: 05:36 Right.

Martin Maron, M...: 05:37 So, take us then maybe through the next step. You got a diagnosis eventually of HCM after again, struggling for a while with misdiagnoses of lung disease. Tell us then, sort of what happened next in your journey.

Shawn Lewis: 05:54 Next was, I went to cardiologist and first visit, I was prescribed the blockers, the [inaudible 00:06:04], the water pills and the [inaudible 00:06:07] and all. The little cocktail, I call it. And then I visited that same doctor again a couple of months later and it just wasn't a fit. And for me, a doctor has to be like a spouse or a mother, or you have to have that relationship with that person. It has to be a fit. It's not a one size fits all, because I'm putting my life in your hands and I think that if I'm not being listened to just as well as I'm not listening to the doctor, it's not going to do either one of us any good. So, I went through several doctors and finally, I found one that was the right fit for me and I was the right fit for him, and we were able to move ahead. And that's where my treatment started and I started seeing some type of improvements.

Albree Tower-Ra...: 06:54 I think that's a very important point. Certainly, as we've been speaking to the variety of symptoms are certainly very diverse. They can be subtle and difficult to interpret as well, in the very beginning. And so, finding a physician that really is a partner in your care is definitely very important. Someone who you feel is able to listen to you and understand where your symptoms are and work with you when you are finding answers or relief for those symptoms, as well.

Shawn Lewis: 07:23 Right. And I was in Georgia at the time. So, that doctor had me go to Atlanta to specialized clinic to do some testing, of course, the stress test and all those different types of tests. I think then, they were trying to... I'm thinking back on it, trying to set me up to try to see if I was qualified for a LVAD, because he had

		mentioned a pump, but that never came, that never happened. I just kept being treated for the cardiomyopathy and the heart failure and then eventually, I moved back to California and started going to Stanford. And one day, I was in a advanced heart failure clinic and I couldn't wait for an appointment, so they sent me straight to Stanford. 65 days later, I came out with a LVAD. I didn't even realize I was that sick because when I went there, it was just an initial appointment, just a first time appointment. So, that's where my journey kind of started, on the healing side.
Martin Maron, M...:	08:33	Maybe we could also just take a step back, because I think you raised a couple other points before that I wanted to also touch on too which is, maybe you could speak a little bit to how effective or not the medicines that you had been recommended to take and perhaps talk at different times were. Did you feel like you had gotten some benefit from those? Did you feel like you were limited by side effects? Were you getting the right information about the drugs from the physicians, in a way that educated you and empowered you, with respect to what they could be doing to help you in this situation? Tell us a little bit about that phase for a minute.
Shawn Lewis:	09:12	Yeah. The drugs did help and the cardiologist was very helpful in making me understand how they worked with this, I call it a disease, with this heart problem. And initially, he was saying that you have to do your part, just like I'm doing my part and just like the drugs is doing their part, which is of course, diet, sodium, as much exercise as you can, but the drugs did help me a lot for as long as they could.
Martin Maron, M...:	09:39	That raises an important point here in HCM is that, we've got a number of very good drug therapies available to help improve symptoms. In your particular situation, which I'm reading a little between the lines, but I think it was nonobstructive hypertrophic cardiomyopathy is what you had. That's the type of cardiomyopathy. We can be in a situation with patients where our current drug regimen starts to lose effectiveness and that's an area part of HCM where we need additional drug development that help patients like you perhaps continue to feel better before having to either undergo an LVAD or maybe avoiding that. I think you provide a great example of the effectiveness of current therapies, but also some of the limitations as well.
Vlad Zaha, MD:	10:28	So maybe just a moment to take another question into the initial evaluation. So, Shawn, do you remember what were the

initial tests that you went through when you met with a cardiologist that you wanted to work with?

Shawn Lewis: 10:43 I went through the echos, the stress tests, there are about five or six. It's so long ago, I'm sorry, I can't remember all of them, but the majority were done in the emergency room and that's how I came to the conclusion that I had the heart failure in the cardiomyopathy. And so, the cardiologist that I eventually stuck with just kept me doing the basic tests that I needed to do and like I said, sent me out to have some more tests done at another hospital, which was either Emory or somewhere in Atlanta.

Vlad Zaha, MD: 11:17 So, and just to follow up on that and then to get some impressions from Dr. Maron and from Dr. Tower-Rader. Do you recall undergoing testing specifically to look for LVOT obstruction or obstruction of your heart?

Shawn Lewis: 11:33 Like I said, they did a cardiac [inaudible 00:11:35]. I don't remember about an LVOT, no.

Martin Maron, M...: 11:40 Vlad is raising it appropriately because patients with hypertrophic cardiomyopathy really have two forms. You can have the one where there is obstruction to blood flow out of the heart from the mitral valve being in the way, called obstructive HCM. And then there's another form called nonobstructive HCM where there's the absence of obstruction. And it's important when a diagnosis of HCM has made, that appropriate testing can be done to put a patient with HCM into the right category of obstructive or nonobstructive, because there's certain therapies that are available only for patients with obstruction and not for those without.

So, it's a really important distinction to make because it affects directly, treatment options and also what patients can expect for their natural history as well. So, that's another kind of important point that probably was done for you. You may not remember it, but it probably was done for you at some point, either at Emory or Stanford, where they looked at that and put you in a, sounds like a nonobstructive category and then the appropriate drug therapy, which then as you said, ultimately stopped working.

Albree Tower-Ra...: 12:55 Yes. Often we look for this either on a resting echocardiogram or you do certain maneuvers, such as a Valsalva maneuver where you bear down. Because sometimes, in a third of patients, we see this at rest or present all the time but in another third of our patients, it's really only with provocative maneuvers that we discover obstruction. So, either with these numerous, like a Valsalva maneuver or with exercise stress testing, as well. So, this was likely looked for at the time of your initial assessment.

Vlad Zaha, MD: 13:27 All right. So, I want to invite here Dr. Maron to comment, maybe on the drugs that are being used for patients with obstructive HCM, with the new developments.

Martin Maron, M...: 13:40 Well, with obstruction to blood flow, the obstructive form of the disease, we've got a number of good drug therapies and we have a number of good, invasive therapies currently, typically in patients that are symptomatic due to the obstructive form of the disease. Symptomatic usually means that patients experience shortness of breath with exertion, or maybe have chest pain or exertional fatigue, the most common symptoms in patients with the obstructive form. We typically will start with AB nodal blocking agents like beta blockers. Occasionally, you can try calcium channel blocker therapies. If those are ineffective or patients have side effects to those, then we can consider selectively in some patients, the drug, disopyramide, which is an anti-arrhythmic drug, but we're using it because it has negative ionotropic properties, which just means that it decreases the force of contractility of the heart that ultimately lowers or decreases the obstruction. That drug can be very effective at making patients feel better by lowering their pressure gradients.

Some patients can have side effects to the drug, but it's generally fairly well tolerated. Although in some situations, in some instances, the efficacy of the drug can wane or decrease over time. For patients that are still symptomatic and frustrated despite those drugs or have side effects to them, then they can become candidates for invasive septal therapies, for which there are really two today, surgical myectomy and a less invasive catheter-based procedure called alcohol septal ablation, which are both very effective, very effective at lowering the outflow gradient and improving symptoms dramatically in patients, with a long-term benefit associated with both of those procedures in mitigating heart failure symptoms.

Small risk to both of those procedures when they're done and performed in expert HCM centers. The recent HCM guidelines recommend that those two procedures be done in highly experienced centers to get the best outcome for patients for

		both myectomy and alcohol ablation. And when done in one of those centers, as I said, there's a substantial improvement, essentially, almost a reversal in symptoms with successful surgery or ablation. So, that's the current treatment strategy for those patients that have symptoms related to obstructive HCM.
Vlad Zaha, MD:	16:27	Thank you, Dr. Maron. And I want to invite now, Dr. Tower-Radar to discuss based on what we learned from Shawn about the nonobstructive HCM that he is suffering from, what medical options are available in that case.
Albree Tower-Ra...:	16:45	As we discussed earlier, the treatments are a little bit different for patients with nonobstructive hypertrophic cardiomyopathy. So, we use some of the same medications, again, the AV nodal agents like beta blockers or calcium channel blockers may be effective. Certainly, if there's evidence that you have extra volume, so the extra fluid like you had discussed before, then we may use diuretics as well to make sure that we're accounting for that, as well. And then if patients remain symptomatic, we really want to make sure again, that we're looking to make sure that we're not missing evidence of obstruction. So as we described, sometimes we don't see this at rest and it's really only with provocation. So, that would be a moment that we would say, let's double check and make sure we're not missing something too. And then for patients who really remain symptomatic despite this, then there may be a role for considering some of the advanced therapies that were suggested for you, like an LVAD or heart transplantation,, in some cases.
Vlad Zaha, MD:	17:47	Thank you, Dr. Tower-Radar. So Shawn, turning back to you now and discussing further, what was your experience as you were telling us at the beginning about your LVAD placement?
Shawn Lewis:	17:59	That was a whole nother experience. As I stated, I went to visit a doctor for the first time in an advanced heart failure clinic and when I left there 65 days later, I had the LVAD. Wasn't expecting it. Life was a blur at that time. Mentally, I wasn't ready. A lot was going on and just didn't know how to handle it. So, mentally, it affected me just as much as it did physically. And after a while, you don't get used to it. I never got used to it. I started having a lot of internal bleeding. I was not ready to be listed for a transplant. In the beginning, they would ask me, "Are you ready to be listed?" No, because I was angry about the whole situation.

		In the meantime, I don't know if I'm going to ahead. In the meantime, my son had the same thing. My father has the same thing. My brother who passed at the age of 24 had the same thing. And all this is just coming to me finding out now that I'm really sick, I'm finding out all these things. So, like I said, I was just angry, mentally. I needed counseling for a chronic illness and I think that's very important for this, for even just the heart failure, the cardiomyopathy, I think people should get some type of counseling, because it's hard to handle being sick all the time. But I made it through and the next phase was the transplant.
Vlad Zaha, MD:	19:46	All right. Thank you for sharing all that really personal information with us. And I would like to invite here our guests to address maybe a couple of points. One is the psychological difficulties of accepting the notion of LVAD implantation and maybe then, the transition to a transplant, as we were going here through a sequence of several steps in the management.
Albree Tower-Ra...:	20:15	I think it's an unusual situation as many of our patients for hypertrophic cardiomyopathy do not necessarily always require an LVAD or heart transplantation but certainly, I think I can comment on the difficulties in coping with a serious diagnosis and how that can be challenging to come to terms with your symptoms and the fact that you're really not feeling well and having to change your perspective in life. And that can be difficult. And certainly, having the support of either friends or family, or your medical providers can be very helpful in navigating that road for you.
Vlad Zaha, MD:	20:53	Excellent. Thank you. And maybe one more question here for our guests is the counseling of the family and exploring the presence of hypertrophic cardiomyopathy in the family members. So, who would like to take this question?
Martin Maron, M...:	21:10	Genetic heart disease. And so, there's important issues related to assessment of family members of affected person with HCM. And so in general, there's a 50% chance that the offspring of affected person with HCM can have HCM. And so we recommend for those reasons, what we call screening, which can take the form of imaging and we recommend that children usually start with a echocardiogram and EKG early. Sometimes it's reasonable to even consider that before puberty. And then certainly no later should that screening start then, typically age 12 or 13, and then echocardiographic screening can be performed usually every year to year and a half through puberty. And then if the echo and testing remains normal in

		that individual, then the echocardiographic screening can be every three to five years. And we usually continue that until usually mid-life or so, and then if things are normal then, it's probably reasonable to stop. So, that's the general assessment of family members, with respect to using cardiac imaging for screening. It can also provide an opportunity to determine if family members are at risk or not of developing HCM through genetic testing. And essentially, that should be performed in conjunction with genetic counselors, hopefully at the institution that this is being done, but essentially, genetic testing results can be used to determine if family members are at risk or not of developing HCM in the future. So, that can also be a useful tool and test to incorporate as well. There are a number of strengths and limitations to that particular strategy that should be discussed with patients before they enter down that genetic testing route but it certainly can be a very powerful test as well in helping to further inform whether patients, family members are at risk or not have HCM. So, those are the two general strategies that one can use.
Vlad Zaha, MD:	23:14	Thank you. And Shawn, turning now to you and we have had a lot of information here that would be useful for a lot of patients. From your perspective, what advice do you have for other patients that might not be aware and then suddenly become aware of new symptoms? What should they discuss with their doctors to reveal that they have HCM?
Shawn Lewis:	23:39	Well, write down everything. I think for a long time, I was in a blur. Like when asked, "What are some of your new symptoms?" Or things like that, I couldn't communicate them. I couldn't... I would just be so frustrated sometimes. So, even if your doctor's appointment is in a month, all month, write down anything that might be a symptom, that you may think that is a symptom, so you can discuss this. The communication with you and your provider is very important.
Vlad Zaha, MD:	24:11	Okay, excellent. I think that sounds good. So, now I would like to turn to our guests, if there is any final advice from each of you, for the patients that are reaching out to physicians with really a need for finding a champion and a good communicator.
Martin Maron, M...:	24:27	A couple of the points that were hit on here that are really important are, finding the right fit. You have got to find the right physician that fits your personality, that you feel comfortable with. There's no question about that, but I think that on top of

that as well, is the idea that particularly in a disease like HCM, it's really important to seek out centers that have particular expertise in the disease. Because of course, if you go to a place that's got a little bit more experience in seeing and taking care of patients with the disease, then the level of interaction in that kind of environment is often different that it may be with less experienced providers. And that makes a huge difference in a disease like hypertrophic cardiomyopathy where seeing a lot of patients and a lot of experience in managing them over time, really counts a lot. And I think Shawn sort of touched on that idea, as well. And so, those are two big, important points, comfort level with who your physician is and also expert level, is really important as well.

Albree Tower-Ra...: 25:31

I would echo that as well. I think in finding a provider that has taken care of patients with hypertrophic cardiomyopathy, you need to think about it before you can look for it. And certainly, that can definitely be one of the things that leads to a delayed diagnosis or in cases, may allow your provider to think about other methods to treat you or to look for other reasons why you may be having symptoms.

Vlad Zaha, MD: 25:56

Thank you, all of you. And in closing, I would like to briefly highlight, again, those couple major points for the physicians to really engage as champions for their patients and to seek the diagnosis beyond the initial appearance and maybe to seek out advice from specialized centers in hypertrophic cardiomyopathy, where there is a lot of experience in managing those patients.

Martin Maron, M...: 26:27

One great resource to help patients, family members find these HCM centers of excellence or expertise is the National HCM Patient Organization, which has a very nice infrastructure for education on the disease, but also can help patients locate providers that have a lot of expertise in the disease as well. And that's something that can easily be found on the internet. The HCMA patient site is a very good resource here.

Vlad Zaha, MD: 27:00

Excellent. I think that's a key point there, something to remember. So, this podcast is part of the American Heart Association HCM Initiative, sponsored by Bristol Myers Squibb. As we were discussing here in closing the session, think about HCMA, think about finding the right fit and seek out the centers with expertise for this disease. I would like to thank now, Shawn, Dr. Martin Maron and Dr. Abree Tower-Radar for joining us, and we hope you enjoyed this podcast. Thank you.

This transcript was exported on Mar 19, 2021 - view latest version here.