Dr. Miguel Leal (00:00):

Hello everyone and welcome to today's podcast promoted by the American Heart Association as part of our educational series on hypertrophic cardiomyopathy, a very important clinical condition that affects an immense number of patients and requires a multidisciplinary approach to its care. I am Dr. Miguel Leal. I am a staff electrophysiologist at the University of Wisconsin-Madison, and I have the privilege to be joined in today's podcast, by a colleague and a patient who will be helping us understand a little bit more about hypertrophic cardiomyopathy and many of its aspects. I have Dr. Robert Fraser with me, and he is a staff cardiologist at the Minneapolis Heart Institute. And I also have Adam Elliot, who is a patient representative. Our goal today will be to go over hypertrophic cardiomyopathy in many of its complex aspects that essentially are part of the day-to-day care of any cardiologist and patient who deal with this condition.

Dr. Miguel Leal (01:01):

We are going to be covering the natural history of the disease and trying to make sure that we focus this on the patient's perspective, tracking patient's symptoms and function, as far as disease progression is concerned, exploring the nuances of obstructive and non-e-obstructive forms of this disease, how it's managed depending on its form, and what is the burden that this disease brings to both patients and clinicians worldwide. So I'm going to start with Dr. Fraser. And Dr. Fraser, I was wondering if you could tell us how you think about your annual hypertrophic cardiomyopathy office visit. Do you follow any specific framework in order to deliver standardized and optimal care to your patients?

Dr. Robert Fraser (01:43):

Thank you for that question Dr. Leal. And I think you bring up a good point that patients with HCM are often seen at least annually in the cardiology office. As far as the framework, I do have a number of things that I try and check off in my head both before I meet the patient, as well as during our visit. I typically review the data with the patient, which would include any testing that was done before we meet. I always evaluate the patient for symptoms and evaluate the patient's sudden cardiac death risk and, or interrogate their device if they have one, typically an ICD, and evaluate the patient for atrial fibrillation. I revisit our screening plan as far as how we're investigating whether or not family members also have the condition. And then I typically close with an element of patient education, which has some recurring themes that I can talk about later. So all in all it involves about five or six objectives for every visit.

Dr. Miguel Leal (02:42):

Thank you Dr. Fraser. That really helps us understand that these visits are carefully planned and there is a method behind the approach to each individual patient, regardless if it's a new patient or a follow-up visit. So I appreciate you sharing that with us. But now I'm going to bring Adam Elliot to the conversation here. And as a patient, Adam, you have a unique perspective because physicians never take their patients for granted and should never do so. But we do have, sometimes dozens or sometimes hundreds of patients with this condition or similar conditions that we see every year. Or it is for you as the patient this is your one and only opportunity to talk to your doctor, oftentimes to share all that has been going on with you. So how do you approach your annual visit with your cardiologist? What goes in your mind as you're preparing for that visit? What do you consider important that you want to share with us here today?

Adam (03:31):

Thank you, Dr. Leal. You really hit the nail on the head with that. Since I was diagnosed in 2009, what I've really learned is trying to make the most out of those visits when I get them. And what I've done to really try to do that and prepare for that is, I know we generally begin with that device interrogation, like Dr. Fraser said, and what I've found is also I've downloaded just a note application on my phone. And that way, generally having my phone with me, I found that that's a way to date and timestamp when I have any particular symptoms. I know that with the device interrogation, if I don't do any recordings myself, that I've notated in my notes, it's more about any triggers that I've had. Again, dates times, what was I doing, anything that might've triggered that. So I try to go in there with almost a hierarchy of any concerns and just go over everything with my physician in the notes and just have a dialogue at that point.

Dr. Miguel Leal (04:37):

Excellent. Thank you for that as well, Adam. So bringing it back to Dr. Fraser, one of the first things you mentioned, doctor, was data review. And I can imagine how much data you have to handle when preparing for the visit with that patient you haven't seen in six or 12 months. So when you talk about that, which studies are you referring to that you're ordering either in anticipation of that clinic visit or shortly after, and how often do you pursue re-evaluation and reassessment of those studies?

Dr. Robert Fraser (05:07):

Sure. So as many of the listeners probably know, hypertrophic cardiomyopathy is a rather heterogeneous disease and it typically requires a personalized approach. It's infrequent that two patients, even within the same family have the same clinical course or require the same care. So I always keep that in mind. I think there's three primary reasons that we order tests as physicians for patients who have hypertrophic cardiomyopathy. The first is for sudden cardiac death risk stratification, to try and clarify whether or not they should be recommended to have a primary prevention ICD. The second is for symptom assessment. And obviously the testing that's ordered is going to be dictated by the symptoms. And then for rhythm assessment, as a part of screening for atrial fibrillation, which is a very common rhythm disturbance that affects on the order of 15 to 25% of patients who have hypertrophic cardiomyopathy.

Dr. Robert Fraser (06:02):

Generally my default approach is all patients with HCM get an ECG at every visit. So it's typically annually. There, I'm just checking rhythm, looking at conduction. Most patients are going to get an echocardiogram and that is a resting echocardiogram, every 12 to 18 months or so. And there we're looking for obstruction. We're looking at LV function, we're looking at the mitral valve and whether or not there's regurgitation. Additionally, some patients, or I should say all patients receive an MRI, that is a cardiac MRI, at their initial evaluation. And then the cardiac MRI is used sparingly based on patient need. And then again, it's dictated by symptoms and how their clinical course progresses over the years that we care for them. So again, default approach is ECG every visit, echocardiogram about every visit or every 12 to 18 months, MRI at first visit and then as needed thereafter.

Dr. Miguel Leal (07:02):

And Dr. Fraser, if I may ask, in terms of genetic testing, genetic evaluation, is this something that once you have ordered initially, do you ever revisit later on or typically once that's done that can be put to rest for a while?

Dr. Robert Fraser (07:16):

Thank you for bringing up that point, as I skipped over genetic testing. Often in my practice, genetic testing is a one-time test to screen for mutations that are known to cause HCM. What we're learning is that as we acquire more data from patients with HCM in these large genetic libraries, that we should maybe consider repeating genetic testing down the road at an interval that's not yet determined because we're learning more and more about HCM genetics, and mutations that we thought maybe were positive years ago, have since been proven to not be causative or vice versa. So currently, that's a little bit of a gray area and generally, I am only doing genetic testing one time for most patients.

Dr. Miguel Leal (08:04):

Perfect. Thank you very much for that. So Adam, you heard Dr. Fraser and you heard the test that he recommends based on current accepted practice guidelines and the standard of care. From your perspective, is that a lot? Is that excessive or is that just about right?

Adam (08:20):

I think it's just about right, Dr. Leal. I noticed from first diagnosed, the echos were about probably every two to three years. And I've noticed that we do do those a little bit more often. And I mentioned earlier about the app in my notes. I have new notes from my last visit with the doctor. And then also I have just a few ongoing notes, I like to call it. And one of those is just to see how that thickening, if there's any more thickening since that last visit. I think having those echos annually helps keep a better read on that. And everything else, as far as genetic testing, MRI, to Dr. Fraser's point, those are things that I had done initially right at my diagnosis.

Dr. Miguel Leal (09:07):

Well, I'm glad to hear that, Adam, because often as physicians, we are not completely aware of the impact that all the tests that we order may have on a patient. So to hear from you that this experience that Dr. Fraser described is considered acceptable by you, it's very reassuring for us as clinicians as well. But Dr. Fraser, we, in the beginning of this podcast, alluded to the burden of disease and how important it is to acknowledge it. And probably the best definition of the psychological burden, if not physical also of hypertrophic cardiomyopathy is the risk of sudden cardiac death. Because it is an unforgiving condition and often leads to horrendous consequences to neurological sequela in some patients when it does not completely result in loss of life. How do you approach this very delicate topic, sudden cardiac death risk prevention?

Dr. Robert Fraser (10:13):

I think this always starts with an element of patient education and trying to help the patient to understand what it is we're talking about when we say sudden cardiac death and that we're implying ventricular arrhythmia that can cause them to die unexpectedly at any time. And really, I approach sudden cardiac death risk stratification as a dynamic and ongoing process or task. It's not really a one-time calculation. Certainly the bulk of it is done at the initial encounter when we establish our relationship between physician and patient. However, I think that it should be revisited annually because patient history and circumstance does change.

Dr. Robert Fraser (10:40):

And so there's two different types of patients that we work with. There's those that do and don't have ICDs. So those that do have qualified based on risk factors for either a primary or secondary prevention

device. And for them, it's a little more straightforward in the sense that we're just doing an annual device interrogation, checking in on device function, making sure they aren't having any complications or issues with the device. Probably more importantly, are those patients who don't have an ICD. And there, I simply revisit what it is we're doing when we talk about screening them for risk factors. We always want to review a family history, whether or not they've had episodes of passing out or syncope or other features of their clinical medical or family history that would warrant upgrading their risk, such that they would qualify for a device. So really in summary, it's all about it being a dynamic process and something that's not done just once, but revisited annually.

Dr. Miguel Leal (11:38):

Thank you. And regarding atrial fibrillation though, when you evaluate patients with this very common atrial tachyarrhythmia, which can cause a stroke, can lead to significant disability, and it's so prevalent in hypertrophic cardiomyopathy patients, how do you approach that risk? How do you assess? Is it the electrocardiogram or do you go beyond that?

Dr. Robert Fraser (11:58):

Yeah, good question. So I think just like we track someone's risk for sudden cardiac death, we track someone's risk for atrial fibrillation. So both the annual ECG, as well as the annual echocardiogram can be helpful. We look at certain features on the ECG, whether or not there's ectopy. With echocardiogram, we're looking at left atrial size and filling pressures. All these things are known to be associated with atrial fibrillation, both in patients with and without hypertrophic cardiomyopathy in there. Certainly especially important in those patients with HCM, we definitely factor in age because we know that the older you get, the more likely you are to develop atrial fibrillation, again, both in patients with and without hypertrophic cardiomyopathy. And then importantly, tracking patient symptoms. So if a patient's describing episodic palpitations or episodic lightheadedness, those are times in which we want to think about having them wear a rhythm monitor, if they are in fact, in normal sinus rhythm at the time of the visit, to try and figure out what their heart rhythm is during these episodes that they're having.

Dr. Miguel Leal (13:03):

Thank you, Dr. Fraser. I'm going to pick up from where Dr. Fraser left in terms of the tracking of symptoms, because we, as physicians, ask questions about things such as shortness of breath, difficulty with physical activities that used to be well tolerated, palpitations or fluttering in the chest. But sometimes we ourselves don't know which symptoms matter more to patients, especially longitudinally. In other words, over time, a symptom may become more or less relevant as the disease is being recognized and treated. So I was hoping you could walk us through your own professional or personal pathway of symptoms here, which is what symptoms matter to you. How do you recognize your disease is compensated or not? And what symptoms are particularly troublesome and alerts you that you should seek medical attention shortly?

Adam (13:53):

Absolutely. And I really appreciate you asking this question because this has been something, I had mentioned that I was diagnosed in 2009. And the reason I highlight that is because I had had symptoms for years prior to that. I would see cardiologists annually as symptoms arose, and used to be fairly active, in fact, pretty intense exercise. And I noticed more symptoms. And those symptoms being, Dr. Leal, that it was the light headedness. It was the palpitations. Even to the point where when I would

walk out in the yard, I would start almost pressing just under my diaphragm, whenever I would feel those irregular beats to get those to go away. And I wore back in the mid 2000s, event monitors, the Holter monitor and everything always came back clean. Had echocardiograms and nothing was really revealed. So it really was a journey to the point where we actually were able to capture atrial flutter in 2008.

Adam (14:52):

Controlled by medication to some extent and then in the fall of 2008 is when I had Afib breakthrough is what they called it. Had an ablation done for Afib toward the end of that year and was still symptomatic. And I think really what was the game changer for me was getting to a practice really where there was a electrophysiologist, someone who specialized in doing the ablation, but also then giving me to the doctors for evaluation for the hypertrophic cardiomyopathy.

Adam (15:25):

So the symptoms really were, it was lightheadedness. I never fully blacked out, but there was times even at the grocery store where I would start feeling that pounding in my chest. I would gray out. There was one time after the ablation where I was at the store and I literally leaned against the wall and my heart was pounding and I came back to, and that's when we followed up, did more tests. And that's when I got the initial diagnosis of the hypertrophic in 2009.

Dr. Miguel Leal (15:58):

Thank you. That was very vivid, an explanation, and really highlights how heterogeneous this disease may be to patients, how different symptoms can present. And also that they're not the same as the disease progresses over time. Dr. Fraser, can you tell us a little bit more about symptom tracking in the two different forms of this disease, which are namely obstructive versus non-obstructive hypertrophic cardiomyopathy?

Dr. Robert Fraser (16:25):

Yes, absolutely. Regarding symptom tracking, I find that again, these patients are often seen about annually unless they develop new symptoms, which would typically warrant a visit in shorter course. And for those with non-obstructive versus obstructive physiology, we tend to think about them a little bit differently when it comes to what could be causing their symptoms. What can make things challenging is that patients with HCM, whether obstructive or non-obstructive, typically describe the same symptoms. That is shortness of breath, with or without chest pain, with or without palpitations. So both types of patients are prone to arrhythmias. And so we always want to keep atrial fibrillation in mind. Both types of patients are prone to sudden cardiac death. And so we screen them the same and it's really about the heart failure where they start to differentiate. So the patients who have non-obstructive disease, we're typically focused on diastolic dysfunction and later in a disease process, possibly systolic dysfunction as a cause for shortness of breath.

Dr. Robert Fraser (17:32):

This can be evaluated often just by echocardiogram, as well as physical exam. In some cases, we'll consider diuretics, but often the mainstay is to start with a beta blocker. Whereas for patients who have obstructive hypertrophic cardiomyopathy, in many ways we consider as classic HCM, the shortness of breath, rather than being mostly due to diastolic dysfunction is actually due to the LVOT obstruction.

Dr. Robert Fraser (17:59):

And in this case, we can evaluate this physiology with echocardiogram. It can also be seen by MRI. But the obstruction is best quantified by echo. And we know that in some cases, we may not detect it by resting echocardiogram. And so if we have patients who are describing shortness of breath, but don't have obstruction at rest, these are the patients who we often ask to do a treadmill or exercise echocardiogram, because we know that patients can manifest obstruction during exercise that they don't manifest at rest. And then we can tailor medications accordingly, which is often either a beta blocker or in some cases verapamil. The most profound instances, referring someone for some kind of septal reduction therapy. So for the most part, these patients describe similar symptoms or manifestations, but the causes can be slightly different depending on whether or not they have obstructive versus non-obstructive forms of hypertrophic cardiomyopathy.

Dr. Miguel Leal (18:57):

So Dr. Fraser, I'm going to switch gears now towards another topic that somewhat connects to the previous conversations we had about genetics, which is family screening. So you have identified your index patient, your pro-band, if you will. And that patient has confirmed diagnosis of hypertrophic cardiomyopathy, be it in its obstructive form or in its non-obstructive form. How do you approach family screening from that moment onwards?

Dr. Robert Fraser (19:24):

Dr. Leal, family screening can be one of the more challenging pieces of caring for a patient with hypertrophic cardiomyopathy. Because as physicians, we have less control over it because those that often matter the most are not in the room, and that is the family members. So often, at my institution, we incorporate a geneticist or a genetic counselor who's comfortable with inherited cardiovascular disease. We ask the patient to speak with this individual. I think it really helps with patient education and understanding of the process as well as informed consent when it comes to genetic testing. And then, like sudden cardiac death risk stratification, it's not a one-time conversation and we revisit the family screening plan at each visit. So whether or not we're using a gene approach or no gene approach, that is if someone's gene positive or gene negative, we often try and simply review patient history, family history, and then do what we can to get in touch with family.

Dr. Robert Fraser (20:27):

There are often barriers. So for instance, maybe siblings, children, parents, they live out of state, or even if they live in state and locally, they may not be able to access our health system due to health plan restrictions. So we try to provide as much patient education as possible and stress the importance regarding family screening, including some written documents that they can pass on, either via email or on paper form, to try and encourage the family members to all get in for screening, at least in the form of an ECG and an echocardiogram. And sometimes, if someone's particularly high risk, say if there's a family history of sudden death at a young age, we may encourage someone to get a cardiac MRI. And that's generally been our approach.

Dr. Miguel Leal (21:11):

Thank you Dr. Fraser. Now, Adam, back to you. You just heard Dr. Fraser's approach to family screening. So did you yourself undergo family screening, meaning did you take the conversation beyond your visit with your doctor and started involving first degree relatives and beyond?

Adam (21:28):

I did Dr. Leal. I had a genetic screening done, probably a year or so after diagnosis. And I think a part of that was, for me, it was like I wanted to know why, or know how, because there was nobody in my family that had had this. And so I kind of wanted, it was the why me, I guess. And so they identified the gene and since then, nobody else in my family has undergone genetic testing. My son that's 14 now, when he was two months old, he had Kawasaki's disease, and it got to the extent that he had a little bit of coronary involvement.

Adam (22:02):

And as it turns out, the hospital that he was at, we started working with the genetic counselor there in addition to his cardiologist. And that same genetic counselor worked with patients to help transition them to the doctor that I see. So it really worked out well that there was a familiarity with the two different practices for my son and for me. And we talked about that with the genetic counselor several times, as far as what are obviously the benefits, I see those, but are there any limitations? It's a family decision. We ended up, my wife and I, deciding maybe to do it whenever they get older, especially if there's symptoms that they might have. My oldest son has been followed over the years, has had echo. So we kind of keep an eye on everything he has had no symptoms. But I think, as he gets older, if he decides to do the testing, we'll involve him in the conversation at that time.

Dr. Miguel Leal (23:01):

Perfect. Thank you for sharing that. Very, very educational for many patients who must be debating if they should or should not involve their families in their own personal diagnosis. We are getting close to the conclusion of today's podcast, but I would like to ask Dr. Fraser one final question today, which has to do with patient education. So we know that it's a complex disease. We just alluded to how there is an obstructive form that sometimes will make people short of breath or sometimes potentially lead them to light headiness and dizziness, because there is not enough cardiac output getting out of the left ventricle through the narrowed outflow tract.

Dr. Miguel Leal (23:38):

But you also have the non-obstructive forms of the disease that may manifest differently. It may show more an arrhythmia type phenotype, where their heart is going fast and sometimes irregularly. And there always the risk of sudden death that permeates both types of presentation. So it seems like patient education is really important as it is for most human conditions, but this one is no exception. So Dr. Fraser, how do you approach that discussion with your patient, understanding that this is something that will take a lot more than one or two clinic visits?

Dr. Robert Fraser (24:10):

Yeah. So I think it's very important to have a patient-centered discussion that is always based on any individual's healthcare literacy and their familiarity with hypertrophic cardiomyopathy. In my experience, there's some recurring highlights that take place during this part of the visit, which is often at the close, and that is some recommendations regarding exercise and its limitations. So we often encourage aerobic over isometric exercises. We generally tell patients with both obstructive and nonobstructive forms to avoid competitive athletics, that they can participate in moderate exercise, but that they should avoid things like burst exertion, so sprinting on a basketball court, for instance. And then also, particularly important for patients who have obstructive forms of HCM, we encourage them to avoid extreme heat and humidity and pay special attention to hydration because we know that

excessive dehydration can lead to further obstruction and some of the downstream consequences like lightheadedness, passing out, potentially instigating arrhythmias.

Dr. Robert Fraser (25:17):

We tell the patients to keep an eye on both signs and symptoms for which they should call into clinic. So whether you have obstructive or non-obstructive forms, it's important to let us know if you develop new shortness of breath or new chest pain or new palpitations. All of these will generally require, at minimum, a conversation if not some downstream testing. We always want to hear about any passing out or syncopal episode. And then I think, very similar to all the patients we see in a cardiology clinic, it's important to revisit things like healthy diet, no smoking or chewing tobacco, and then healthy alcohol consumption, which is relevant to everyone that we see. So it's a little bit of a grab bag. It's certainly tailored to any given patient, but those are just some of the highlights that I find myself covering over and over again.

Dr. Miguel Leal (26:06):

Thank you Dr. Fraser. That is the perfect summary, I think, to today's discussion. And Adam, you've been an interesting part of this conversation from the beginning. And I, again, cannot emphasize how important it is to have your perspective. If you're a patient who is listening to this podcast, do you think that this education agenda, do you think that having a physician invested in trying to teach you, one step at a time, about your disease align with your wishes and is that enough? What else should physicians do to keep in mind that patient education is paramount in managing this disease?

Adam (26:42):

I appreciate that very much. I think the biggest thing is for me, it's always wanting to learn more and that's using the resources that are out there. When I was first diagnosed, I was given some information that day for the hypertrophic. And to Dr. Fraser's point, we talked about some of the limitations, the things to be aware of, the hydration and the ways that it's affected how I do what I do, when I go on, if I go on scout camp outs with both my boys, it's typically avoid the hot weather, those kinds of things. I try to pick and choose when I do what I do. But I think the resources that are available, one time I just had the opportunity to talk with a patient before I had my ICD placed. And just having somebody else to talk with, that's why I think this podcast is so valuable because I would have loved to have something like this when I was first diagnosed as a resource and just to have everything in one place, on demand.

Dr. Miguel Leal (27:41):

Thank you, Adam. This concludes today's podcast, as part of our educational series on hypertrophic cardiomyopathy. I'd like to thank Dr. Robert Fraser and Adam Elliot for their participation today. And I hope you'll have the opportunity to join us for future podcasts on this very important topic. Thank you. Sponsored by MyoKardia.