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Hypertrophic Cardiomyopathy: Challenges in Diagnosis & Management

Announcer:
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This medical industry feature, titled "Hypertrophic Cardiomyopathy: Challenges in the Diagnosis & Management" is provided in partnership with Bristol Myers Squibb. This program is intended for physicians.

Here's your host, Dr. Matt Birnholz.

Dr. Birnholz:
Hypertrophic cardiomyopathy, or HCM, is the most common inherited genetic heart disease, affecting approximately 1 in 500 or more patients in the U.S. However, it has been estimated that many patients remain undiagnosed.

This is ReachMD and I'm Dr. Matt Birnholz.

Joining me to discuss the diagnosis and current approach to management of HCM is Dr. Andrew Wang. Dr. Wang is Vice Chief for Clinical Affairs for Duke University's Division of Cardiology and directs the Duke Hypertrophic Cardiomyopathy Clinic. He's also a paid consultant for Bristol Myers Squibb. Dr. Wang, thanks for being here, today.

Dr. Wang:
Thank you for having me here, Matt.

Dr. Birnholz:
So, let's dive right in, Dr. Wang. To start, can you give us some more background on HCM and underdiagnosis and why it's such a problem?

Dr. Wang:
Yes, Matt. HCM as you mentioned is known to affect about 1 in 500 persons, that's across all sexes, ethnic groups, geographies and yet as you mentioned it goes undiagnosed.

The reasons for the underdiagnosis is multiple. Some patients are asymptomatic throughout their lifetime and obviously would not seek medical care. The spectrum of symptoms and their severity are very wide across patients with hypertrophic cardiomyopathy. Some people have only palpitations, other people have some occasional chest discomfort, other people have more progressive symptoms, shortness of breath, passing out spells. So, there's a very wide spectrum and many of those symptoms are found in other heart conditions, other more common heart conditions. And so hypertrophic cardiomyopathy may not be the top of everyone's mind when they're seeing a patient with palpitations or some dyspnea on exertion.

The last one is that patients, often times, adjust their level of activity or their lifestyle to their symptoms of hypertrophic cardiomyopathy. I've heard many patients attribute their "slowing down to age" when it really is their cardiac condition that is causing them not to be able to perform their activities.

Dr. Birnholz:
Interesting. And with that in mind, Dr. Wang, can you just elaborate on some of the consequences of undiagnosed or untreated HCM?

Dr. Wang:
The most feared consequence is sudden cardiac death. I think that we have heard stories, often times, in young athletes. They're

competing in a very elite or high level and suddenly they collapse and are found to have hypertrophic cardiomyopathy. Fortunately, that does not affect the majority of patients with hypertrophic cardiomyopathy.

The other consequence is progressive heart failure symptoms. For example, patients who have hypertrophic cardiomyopathy may have the gradual onset of symptoms of shortness of breath, but this can progress over the years to very severely limiting shortness of breath. But in addition, it's important to recognize that HCM is a progressive disease and that patients may begin with mild symptoms.

Dr. Birnholz:
So, Dr. Wang, given these impacts in the context of underdiagnosed disease, can you walk us through some criteria we should keep in mind for diagnosing HCM?

Dr. Wang:
There are many findings and symptoms that should raise the question of hypertrophic cardiomyopathy to the healthcare provider. It may be a abnormal systolic heart murmur on physical exam with some dynamic features that change with maneuvers. Patients sometimes will have a screening or routine electrocardiogram that shows evidence of left ventricular hypertrophy or repolarization abnormalities that suggest hypertrophic cardiomyopathy. Patients will sometimes report non-specific symptoms such as palpitations, occasional lightheadedness, fatigue, mild shortness of breath that's attributed to some other condition, for example, like asthma. And so, keeping a index of suspicion for hypertrophic cardiomyopathy in these settings is important. Lastly, I would say in a patient presenting with a history in the family of someone with a cardiomyopathy or often described in the lay descriptions as "an enlarged heart" or someone who has died suddenly should also help to increase the suspicion for hypertrophic cardiomyopathy.

Dr. Birnholz:
Excellent. And can you also talk to the role of cardiac imaging and its place in diagnosing HCM?

Dr. Wang:
Cardiac imaging is the critical and most important diagnostic test and step to find and identify hypertrophic cardiomyopathy. The echocardiogram, transthoracic echocardiogram, usually serves as the diagnostic test of choice for hypertrophic cardiomyopathy. In addition to echocardiography, cardiac MRI is helpful for diagnosis confirmation, particularly in cases where there's some uncertainty based on the echocardiographic findings and can help to show other features that may help manage the patient with hypertrophic cardiomyopathy.

Dr. Birnholz:
Really insightful, Dr. Wang, thank you.

For those just joining us, this is ReachMD. I'm Dr. Matt Birnholz and I'm speaking with Dr. Andrew Wang about hypertrophic cardiomyopathy

Now, Dr. Wang, we spoke about HCM diagnosis but I wanna shift gears and turn to how this disease is managed. So, let's talk about the management strategies. What can you tell us about the current management strategies for HCM?

Dr. Wang:
It's important to note before I begin that most of the pharmacologic options included in the guideline recommendations are not FDA approved for hypertrophic cardiomyopathy. The drugs that are used to manage hypertrophic cardiomyopathy are used for many other cardiovascular conditions but are not specifically labeled or indicated for treating hypertrophic cardiomyopathy.

I think it's important to recognize there are two main subtypes of hypertrophic cardiomyopathy: those that have left ventricular outflow tract obstruction, and those that have non-obstructive hypertrophic cardiomyopathy. Some patients, importantly, have no significant resting left ventricular outflow tract obstruction, but do have a significant provocable obstruction, either with Valsalva maneuver or with exercise testing.

Current management strategies for caring for the patient with hypertrophic cardiomyopathy are based on the 2020 American Heart Association, American College of Cardiology guideline for the diagnosis and treatment of patients with HCM. The guideline is very extensive and covers the range of issues in caring for and taking care of a patient with hypertrophic cardiomyopathy; from diagnosis confirmation to management of symptoms, to risk stratification for sudden cardiac death, to managing other known complications of hypertrophic cardiomyopathy and then also, for the important issue of family screening and genetic testing.

For the patient that has symptoms with obstructive or non-obstructive hypertrophic cardiomyopathy, the first line therapy is beta blockers and if those are not tolerated or ineffective for managing symptoms the other treatment would be non-dihydropyridine calcium channel blockers as the next line of therapy. If trials of beta blocker therapies or calcium channel blockers are ineffective and symptoms continue to persist in patients with obstructive HCM, another option would be to try disopyramide in combination with one of the other drugs such

as beta blockers and calcium channel blockers to manage symptoms.

Finally, in patients that have refractory symptoms despite available pharmacologic therapies, septal reduction therapies, such as surgical myectomy and alcohol septal ablation can be used to reduce left ventricular alpha tract obstruction.

Dr. Birnholz:
Well, that's a great overview, Dr. Wang. And I think it opens up a natural extension question for you, which we can use to close the program. And that's, based on your experience, what changes would you like to see?

Dr. Wang:
The current pharmacologic treatment options such as beta blockers and calcium channel blockers are typically indicated for other cardiovascular conditions. And so, there is a treatment gap currently for managing these patients pharmacologically.

Dr. Birnholz:
Well, with those take-aways in mind, I very much wanna thank my guest, Dr. Andrew Wang for helping us better understand hypertrophic cardiomyopathy. Dr. Wang, it was great speaking with you today. Thanks so much for joining us.

Dr. Wang:
Thanks for the time today to talk about this important cardiovascular disease.

Announcer:
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