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HCM vs HFpEF: Avoiding Common Diagnostic Pitfalls

**Announcer:**
You're listening to GLC on ReachMD. This activity, titled **'H-C-M versus Hef-pef: Avoiding Common Diagnostic Pitfalls,'** is provided by Global Learning Collaborative.

**Dr. Masri:**
Hello, I'm Dr. Ahmad Masri. Hypertrophic cardiomyopathy is often hiding in plain sight. It's frequently misdiagnosed as HFpEF because both conditions can present with preserved ejection fraction and have similar symptoms, such as dyspnea or exercise intolerance. But these are 2 fundamentally different diseases with very different implications for management and risk, including sudden cardiac risk in hypertrophic cardiomyopathy and including cascade family screening and evaluation. And so today we'll explore why this confusion happens, why it matters, and how clinicians can more confidently distinguish between these 2 conditions in practice.

The first thing we'll touch base on is why does HCM get misdiagnosed as HFpEF? I think the primary reason is that why is the patient coming to you? They're short of breath, they're fatigued, they have reduced exercise capacity, or they're retaining water in their legs or things like that. And so typically an EKG leads to an echo, which leads to something called either you have preserved ejection fraction or you have reduced ejection fraction. And very commonly patients have preserved ejection fraction, and so that's where this chronic diastolic heart failure or heart failure with preserved ejection fraction comes into play.

But the reality is and the difficulty is that there's a lot of shared risk factors between hypertrophic cardiomyopathy and HFpEF that includes hypertension, obesity, sleep apnea, alcohol, smoking, and metabolic syndrome. And as such, we over rely, really, on echocardiography with this single parameter of ejection fraction to say that someone has disease X or disease Y. And on top of that, there is general underappreciation of what HCM looks like on imaging, on echocardiography, in the absence—especially in the absence—of very clear LVOT or left ventricular outflow tract gradient. And so the bias is that because of how common HFpEF is, the bias is just to say that everybody has HFpEF, and the bias is to look at anyone who has metabolic syndrome and who's obese and say you have HFpEF.

And so why is this important? It's important because management is different. So first of all, if someone has HCM, they require further workup, and they require family, as well, evaluation, potentially genetic evaluation. And right now, we have appropriate management options for these patients that can normalize in a lot of them how they feel from day to day, while in HFpEF it tends to be a little bit more of a difficult journey; you have to pick and choose and try to find the right therapies for them.

And so the other piece of it is that we believe in general that having specialty assessment of patients with HCM is useful and leads to better outcome. And as such, identifying these patients will be useful to any practice, not just in referral or academic centers, but also in the community settings and even in the primary care settings.

And there are clues. There are lots of clinical clues that you can use to raise your suspicion for HCM. I think the most important piece to keep in mind is that you always need to explain left ventricular hypertrophy. So if you just start there and say that any patient that you see and they have left ventricular hypertrophy, especially when it's above 13-14 mm, you want to strive to explain that. Do they have hypertension, metabolic syndrome? Is it sufficient? So all of these things can help you really isolate what's LVH related to these things versus what's out of proportion.

Someone who tells you, “For 20 years I have never seen my blood pressure uncontrolled,” and they have normal kidney function, that septum of 18 mm is very unlikely to be related to hypertension.

And the other piece of it is there is discordance of how they feel. And so that patient who comes to you, they could have severe left ventricular hypertrophy while they're symptomatically doing okay, and they're not swollen, and they're not in heart failure. That's something to keep in mind.

And so back to the story of differentiation, as you can tell from all this talk so far, a lot of it has to do with clinical symptoms but also with imaging. Now, clinical symptoms are important because patients with HCM tend to not swell up, while patients with heart failure with preserved ejection fraction, they tend to have lots of fluctuations and cycles of congestion, decongestion. They need diuretics and whatnot. HCM at rest usually does not lead in early disease or intermediate disease for that to happen. Usually patients are short of breath on ambulation, but you examine them and you're like, you look great, you don't have really any swelling or any volume overload here, so I'm not so sure what's going on. So that usually is a clue.

Combine that with having left ventricular hypertrophy that specifically is asymmetric. And when we talk about asymmetry, it's more or less you have a thickened septum while your lateral wall is really not that thick. If your lateral wall is thick, even if it's out of proportion to the septum, this could still be not HCM or could be HCM. You have to work these patients up a little bit more. But if you have a very thin lateral wall and you have the septum to be very thick, that's a big red flag.

The other piece of it is left ventricular outflow tract obstruction, and this is where a lot of the diagnostic errors also happen. At rest, when we define it above 30 mmHg or with provocation above 50 usually, you have that absence at rest in half the patients of obstructive HCM. So you need to be provoking these patients so that you can see what happens. So that's an effective Valsalva that you have to invest in your clinic, in your echo lab. You have to invest in sonographer training to do adequate Valsalva.

Diastolic function measures are not super useful, but ejection fraction is. Patients with HCM tend to run on the higher side of EF, so 60s and above, hypercontractile. Patients with HFpEF tend to run on the lower end of the spectrum, 50s to 60s. And if you have someone with severe hypercontractility and severe LVH, that's a good, good clue. Also LV cavity size. If you have, for example, a man who's big-sized and they have an EF of 75% with a very small cavity size, that's again a red flag that this could be hypertrophic cardiomyopathy.

And then finally, this should lead you frequently to go for MRI. And if you go to MRI, you can tell and see what's going on at the structural level, functional level, as well as evaluate the patterns of left ventricular late gadolinium enhancement, which allows you to differentiate sometimes—not all the time—but sometimes can allow you to differentiate between HFpEF and HCM and, more importantly, pick up on other diseases that cause LVH, such as amyloid and Fabry disease, which have their own independent treatments as well.

All right. So why does it matter to differentiate for obstructive versus nonobstructive HCM? Because it has implications on therapy. And these implications on therapy have to do with the fact that we have very effective treatments nowadays—cardiac myosin inhibitors, septal reduction therapy—that can relieve that LVOT obstruction and provide patients with almost normal—normal way of life and feeling from day-to-day basis. So that's something really important.

For nonobstructive HCM, it is not that there is nothing whatsoever to do. There are some things you can try, but also there are clinical trials that are evaluating this. So this will become even more important in the future to identify who has nonobstructive HCM versus HFpEF.

And so moving on to some practical steps that we can avoid some of these diagnostic pitfalls. Again, my favorite is to try and explain all left ventricular hypertrophy. If you have discordance of NT-proBNP and symptoms, that's a red flag for HCM or potentially even amyloid if the discrepancy is pretty high. So someone comes to you, they're pretty good, they're NYHA class II, even barely II, maybe I, but their NT-proBNP is 1000, that's when you should look more and what's going on. This is a myopathic process that is likely happening in the absence of congestion. In HFpEF, elevated NT-proBNP follows these congestion cycles pretty frequently.

Also arrhythmias. Arrhythmias are common in both conditions, including atrial fibrillation. So that's on its own might not be a good clue for you. But ultimately if you employ a systematic strategy in your clinic trying to evaluate patients who have left ventricular hypertrophy

and those who have LVOT obstruction and those who have hypercontractile ventricle with very high LVEF above 65%-70%, I think you can capture the majority of the patients.

What can you do tomorrow? I think a few things. One, discuss with your echo lab standardizing and doing better with Valsalva evaluation in anyone who has left ventricular hypertrophy. And then two, for anyone who has heart failure symptoms but does not have frank signs and symptoms of HFpEF or heart failure and congestion-decongestion cycles, you should obtain a cardiac MRI to help you with this evaluation. And the third step, don't forget offering for genetic testing if you suspect hypertrophic cardiomyopathy because you can help lots of families in identifying these conditions and diseases through cascade testing. And so this is all to just say that if you identify these patients, you have therapeutic options for them, and that is really, really important.

And so finally, to wrap us up, not every patient who has preserved ejection fraction and heart failure symptom is HFpEF. There is a lot of other diseases that you need to rule out and make HFpEF a diagnosis of exclusion. Once you have excluded all the other diseases that can particularly cause the same phenotype, you will have a different approach to management, risk assessment, and you will improve your patients' outcomes.

That's all we have for you today. Thank you for joining me.

**Announcer:**
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