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Exploring Patient Perspectives on Genetic Testing for ATTR-CM

Announcer:

You're listening to *On the Frontlines of ATTR-CM* on ReachMD. Here's your host, Dr. Shelina Ramnarine.

Dr. Ramnarine:

This is *On the Frontlines of ATTR-CM* on ReachMD. I'm Dr. Shelina Ramnarine, and joining me to discuss his recent study, which looked at patient perspectives on genetic testing for transthyretin amyloid cardiomyopathy, or ATTR-CM, is Dr. Quan Bui. He's a cardiologist and Assistant Professor of Medicine at UC San Diego Health. Dr. Bui, thanks for being here today.

Dr. Bui:

It's my pleasure.

Dr. Ramnarine:

So to set the stage for our discussion, Dr. Bui, could you tell us a little bit about your study and what motivated you to interview patients with ATTR-CM and their at-risk family members?

Dr. Bui:

Yes, so genetic testing is something that's really near and dear to my heart, taking care of a lot of patients with genetic cardiomyopathies. And so it all kind of began at the bedside, where I thought that we were not utilizing genetic testing enough in our patients. And so I had lots of patients come to me with idiopathic cardiomyopathy, and never really had other people think about what could be the etiology of their cardiomyopathy. And so I think those are opportunities to figure out more and understand that they might have a genetic cause of their disease. And so I basically used my expertise in cardiac amyloidosis to understand what the barriers to genetic testing are in that patient population. And I've found out a lot of things that were very interesting, and we'll probably discuss more about it, but it really began at the bedside, learning more about why are we not doing enough genetic testing in our patients.

Dr. Ramnarine:

And from my understanding, there were four key themes that emerged from your research, so I'd like to go through each of them in depth. The first was how many patients relied on trusted providers to guide their decision-making. Could you tell us why provider recommendations are such a pivotal factor and how we can better equip non-genetic clinicians to play that role?

Dr. Bui:

Yeah, that was probably the biggest finding of our study: that patients really relied on their providers to guide them through this process, right? The patients themselves really don't know much about genetic testing, and so if the providers brought it up and suggested genetic testing, they had complete faith in their team to proceed with that recommendation. It's almost kind of blind trust. It's kind of interesting, where patients say, "Yeah, whatever you want to do to me, do it," right? But I found out that patients, even though they want to follow their provider's recommendation, they also want to know what the pros are and what the cons are of genetic testing as well. And so having the provider go through that information with them, they found was very important.

Dr. Ramnarine:

And another theme was an overall positive attitude towards genetic testing. Why do you think patients felt that way, and how did this perspective factor into their decision-making?

Dr. Bui:

I think that patients, overall, see genetic testing as getting more information into their disease, right? There's a big focus on whether this genetic testing information would help change their management or change the way their provider looks at their disease prognosis. And so the overall theme was that patients want to know more information. That was one thing. And then the other thing that was also important to them was, how do they protect their family members? If this is genetic, could this be helpful with their siblings? With their kids? And could that allow for earlier diagnosis of potential amyloidosis and earlier treatment? And so I think those were the two things that resonated with patients around genetic testing: that increased knowledge, as well as how this could potentially help manage their kids and their family and prevent any complications down the road.

Dr. Ramnarine:
For those just tuning in, you're listening to *On the Frontlines of ATTR-CM* on ReachMD. I'm Dr. Shelina Ramnarine, and I'm speaking with Dr. Quan Bui about patient experiences with genetic testing in ATTR-CM.

So, Dr. Bui, let's continue our discussion of the four key themes you uncovered. Your next finding was that, despite positive attitudes overall, the emotional impact of genetic testing was significant. With that being said, what kind of psychological stress did patients report, and how might care teams better help them navigate these decisions?

Dr. Bui:
I think that, overall, patients really found genetic testing to be positive, as you mentioned, but there are certain things that they want to know about and also be reassured about. So one thing was anxiety around the genetic testing results and knowing what to expect. And so some of the patients described they didn't really know how long it would take for the genetic testing to come back, so even explaining to them what to expect and how long it may take that can help out with some of that anxiety.

Obviously, especially with asymptomatic family members who are getting cascade tested, they're worried that they may have the same fate as their family members with amyloidosis, and so there's a lot of anxiety around that too, knowing that it could be a 50/50 chance for them to have a positive genetic test. And so providers can provide reassurance and say, "Hey, even if we do find a positive test, uncertain results, or a negative test, we're here to support you," right? And we have a great team around diagnosing and managing amyloidosis and we have support from genetic counselors and genetic specialists that I think patients felt reassured by. I think for the patients that have a lot of anxiety around genetic testing, non-genetics providers should and may consider referral to genetic counselors who are better equipped with managing some of those anxieties and emotionally provoking experiences. So knowing that you have genetic support or genetic counselors who can be available would be helpful to also mention to patients.

Dr. Ramnarine:
So the final theme that emerged was that there were gaps in how pretest counseling and informed consent was handled. What did patients express about this, and how can that be improved?

Dr. Bui:
This is actually something that's really important to me, really making sure that patients understand what the potential risks that are associated with genetic testing are. And this is especially relevant for patients who are asymptomatic and who are getting maybe cascade testing if a separate family member was positive. And so it's important to disclose and go over potential insurance ramifications. So, luckily, there was a law passed in 2008 that says that, for the most part, the workplace and health insurance cannot discriminate against you for having an underlying genetic condition, but for life insurance, disability insurance, or those types of things, if they were to come across that information, there could be some potential ramifications. So it's important to disclose that information. It's less relevant for patients who have an underlying amyloid cardiomyopathy, right? But patients still appreciated the knowledge. They wanted to know that. And even though it wouldn't change their overall decision to get genetic testing, they still found that it was important information to know as part of the decision to partake in genetic testing.

Dr. Ramnarine:
So before we come to the end of our program, Dr. Bui, do you have any final insights on how we can better support patients considering genetic testing for ATTR-CM?

Dr. Bui:
Yeah, I think that genetic testing is viewed very positively by patients, at least in this study. I have to say, it's a small study, but it gives us a glimpse into what patients think about genetic testing. I think us as cardiology providers need to embrace genetic testing and take it upon ourselves to discuss these important things with our patients so that it can potentially alter the treatment of the patient in front of us, but also identify subclinically affected family members who may benefit from additional screening and potentially initiation of therapy to prevent disease down the road. And so it is really our responsibility as cardiologists and as cardiovascular providers to be the champions of genetic testing.

Dr. Ramnarine:
So with those key takeaways in mind, I want to thank my guest, Dr. Quan Bui, for joining me to discuss how we can improve the genetic testing process for ATTR-CM patients and their at-risk family members. Dr. Bui, it was great to have you on the program.

Dr. Bui:
Thank you so much. I appreciate it.

Announcer:
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