Transcript Details
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Overcoming Challenges in ATTR-CM Care: Best Practices for Accurate Diagnosis

Announcer :
You're listening to *Heart Matters* on ReachMD. On this episode, we'll hear from Dr. Michelle Kittleson, who's the Director of Postgraduate Education in Heart Failure and Transplantation and Professor of Medicine at the Cedars-Sinai Smidt Heart Institute in Los Angeles. She'll be discussing the role of comprehensive multidisciplinary care in transthyretin cardiac amyloidosis, or ATTR-CM. Let's hear from Dr. Kittleson now.

Dr. Kittleson:
The key challenges in diagnosing ATTR-CM, or transthyretin cardiac amyloidosis, include, number one, a high index of suspicion in the face of clinical clues. This is not a diagnosis that will jump out at you on routine testing. You have to think about it to order the right test to diagnose it. If you have an older patient with dyspnea, edema, increased left ventricular wall thickness, and ejection fraction somewhere around higher than 40 percent—so that typical heart failure with mildly reduced or preserved ejection fraction phenotype—think about this diagnosis. Other supporting findings would be a history of bilateral carpal tunnel syndrome, spinal stenosis, or peripheral neuropathy since the transthyretin amyloid protein infiltrates other parts of the body. So have a high suspicion for that picture to order the appropriate testing.

And that leads us to the second diagnostic challenge, which is ordering the right tests and interpreting them in the right way. So the two most common forms of cardiac amyloidosis come from immunoglobulin light chains—that's AL amyloidosis, a plasma cell dyscrasia along the spectrum from MGUS to multiple myeloma, and the TTR cardiac amyloidosis, which comes from the transthyretin or TTR protein in which there are two forms. One is the variant form caused by a mutation in the gene, and the second is the wild-type form. You don't want to miss a diagnosis of AL cardiac amyloidosis, which can be a medical emergency warranting immediate referral to a hematologist for diagnosis and plasma cell-directed therapy.

So how are you not going to miss the diagnosis? How are you going to tell the two conditions apart? Number one, you must order a monoclonal protein screen, which must include serum immunofixation electrophoresis, urine immunofixation electrophoresis, and serum free light chains. Protein electrophoresis is not sensitive enough—it must be immunofixation electrophoresis. If there's a monoclonal protein identified from the serum IFE, urine IFE, or serum free light chains, that is an immediate consultation to a hematologist and to perform a biopsy because AL cardiac amyloidosis requires a biopsy for diagnosis. It may start with something that's less invasive, like a fat pad, which is a surrogate site, but that lacks adequate sensitivity. So if negative, you will then proceed to biopsy the affected organ, typically heart or kidney. But if the monoclonal protein screen is negative, you will breathe a deep sigh of relief and turn to your bone scintigraphy scan—most commonly technetium pyrophosphate scan. If the technetium pyrophosphate scan is positive and the monoclonal protein screen is negative, this is diagnostic of TTR cardiac amyloidosis with no need for a tissue biopsy. However, 10 to 20 percent of patients with AL cardiac amyloidosis can have a false positive technetium pyrophosphate scan, which is why that scan can never be interpreted outside the context of a negative monoclonal protein screen. But if your monoclonal protein screen is negative and your technetium scan is positive, the patient has TTR cardiac amyloidosis, and you will then perform genetic testing to assess wild type versus variant form, which will be useful for some therapeutics, as well as cascade screening of first-degree relatives.

It's very important to remember the role of multidisciplinary care in patients with cardiac amyloidosis because the amyloid protein just doesn't go to the heart. It can go to other places as well. Unique to AL cardiac amyloidosis is a nephrotic syndrome or nephropathy, and both can cause gastrointestinal manifestations, types of malabsorption, and malnutrition, which can be related to infiltration or the neurological involvement.

There are clues to diagnosis. An older patient with bilateral carpal tunnel syndrome should be a red flag to consider that this diagnosis is from cardiac amyloidosis. One approach that is gaining favor from many orthopedic surgeons is to send the specimens from carpal tunnel surgery or spinal stenosis surgery for evaluation by a pathologist for Congo red staining and subtyping for amyloid involvement as this could lead to earlier diagnosis of cardiac amyloidosis.

Announcer :
That was Dr. Michelle Kittleson discussing multidisciplinary management in patients with ATTR-CM. To access this and other episodes in our series, visit *Heart Matters* on ReachMD.com, where you can Be Part of the Knowledge. Thanks for listening!