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The Multiple Faces of ATTRv-PN

Announcer:

Welcome to CME on ReachMD. This episode is part of our MinuteCE curriculum.

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Dr. Waddington-Cruz:

So this is CME on ReachMD, and I am Márcia Waddington-Cruz. With me today is Dr. Sami Khella.

So, Sami, can you walk us through a case of ATTRv polyneuropathy? We are especially interested in the variations in clinical presentation and the different identifying signs and symptoms. So please, Sami.

Dr. Khella:

Thank you so much, Márcia. Let me tell you about J.R. He's a 74-year-old gentleman who has a long history of diabetes that has been moderately well controlled on insulin. And then he developed the sudden onset of numbness and tingling in both upper extremities that he had not had previously. In the past, he had had some numbness and tingling in his feet that had been stable for a long time, but these, after he developed the numbness and tingling in his upper extremities, also started to progress in his lower extremities. At the same time, he developed atrial fibrillation and was being seen by a cardiologist for heart failure that was new in onset. His numbness and tingling continued to progress, and it was associated with lumbar spine pain that radiated into both lower extremities. But he was also getting weak at the same time, having trouble going up and down steps and raising his arms above his head to wash his face. The symptoms continued to progress, and he also, at the same time, was losing weight, as I said, developed diarrhea alternating with constipation that was quite severe and prevented him from going outside of the house. He was having trouble walking with gait instability and was progressively getting weaker to the point that now, within about 6 months, he had to use a wheelchair.

He underwent lumbar spine surgery for a presumed lumbar spine stenosis, but this did not help his weakness in any way. At the same time, he was noted by his cardiologist to have progression of his heart failure to the point where he was developing thickening of the heart wall on echocardiography, and the atrial fibrillation was difficult to control as well. So it turned out that this patient had a brother who had developed similar symptoms about several years prior and then died of unknown causes about a year prior to the presentation. So this is a difficult and a complex patient that I recently saw. Hopefully this was helpful.

Dr. Waddington-Cruz:

So my takeaway message from the case that was presented is that you should always think about the possibility of having a diagnosis of ATTRv polyneuropathy and should, whenever there is a red flag, like autonomic dysfunction, whenever you have a family history, or if the family history is not obvious at the beginning, as he mentioned, that only after the brother came with the with the same presentation, and whenever there is heart hypertrophy or conduction of abnormalities, and several other signs, red flags that we'll talk about later, then just think about the possibility the patient is progressing, is not responding well to treatment. So just think about and look for the diagnosis and for the possibility, because it's a treatable disease.

Announcer:

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