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ATTRv-PN: A Complicated Disease With a Poor Prognosis

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

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Dr. Berk:
This is CME on ReachMD and I'm Dr. John Berk. Here with me today is Dr. Sami Khella, a professor of neurology, and we're going to discuss transthyretin amyloidosis with polyneuropathy, which is a complex, debilitating disease that impacts patient quality of life and can be a burden on family caregivers.

Sami, can you give us some insight on the history of amyloid polyneuropathy and how this disease affects not just the patient but the families as well?

Dr. Khella:
Sure, John, thank you. Amyloid polyneuropathy is a really variable disease and a tricky one to diagnose in the beginning because it looks like the very common sensory neuropathy that most neurologists confront in their clinical practice. But this is really a systemic illness and some of the special features of this neuropathy, John, as you well know are the weakness that it causes. And a lot of times people think that they have a patient with CIDP [chronic inflammatory demyelinating polyneuropathy], and this CIDP patient is not improving on the current therapy. And characteristic of CIDP is a proximal as well as a distal weakness, and weakness really is the feature of hereditary amyloid polyneuropathy. But the weakness is proximal as well as distal, and it looks just like CIDP. But very different from CIDP is the autonomic symptoms that these patients have. They get diarrhea; they get constipation; they have lightheadedness. The men have early-onset erectile dysfunction. But I want to highlight something about this neuropathy, and I'm going to just contradict myself completely by saying that the weakness may not actually be present in all cases of hereditary amyloid polyneuropathy. And the reason is that, in the United States, the V122I variant, which is the variant that affects almost exclusively African Americans, is the most common variant that the neurologist will probably see. But that variant doesn't have much in terms of weakness. So unlike the more common variants, the V30M, which is the most prevalent variant in the world, V122I doesn't have a lot of weakness associated with it.

Now, the second most common, and another one that, especially on the East and West Coast, neurologist will come in contact with, is the T60 variant. And that one is often a mixed phenotype, a cardiomyopathy peripheral neuropathy phenotype, and that can have a significant amount of weakness.

I don't know. Is this your experience as well, John?

Dr. Berk:
Yeah, I think that's true. I think one of the perplexing things, not being a neurologist, is that despite the fact there have been therapeutics available over the past 5 to, actually, 10 years, it appears to me like we're not doing a very good job at identifying people with neuropathy in an early fashion. What's your take on that?

Dr. Khella:
I think the thing that we have to, as neurologists, really focus on is that this is a progressive illness. So this neuropathy, unlike diabetic neuropathy, unlike the neuropathy from B12 or even chemo therapeutic drugs, the neuropathy of amyloidosis is progressive. And so I think that when a patient is seen with early-onset small-fiber or just pure sensory neuropathy, I think it behooves the practitioner to see that patient in follow-up in 6 months, in 12 months, and that's generally my practice, to see them 2 or 3 times again over a fairly substantial period of time to be absolutely sure that this neuropathy is not progressive. And I think this is where people make a mistake sometimes in that they will see a patient once and then not follow up with that.

Dr. Berk:
And do we have sufficient genetic testing among neurologists?

Dr. Khella:
Yeah, I think it's getting better. I think the cardiologists are doing a much better job at getting genetic testing. There may be some hesitation on the part of neurologists. I'm not really sure exactly all the reasons why neurologists don't test. But of course, you know, the pharmaceutical companies now have free testing, so you can order just a TTR gene if you're looking for hereditary amyloidosis.

Dr. Berk:
Well, unfortunately, our time is up, but that was a great discussion. I learn something every time I speak to you, Sami. Thanks a lot.

Announcer:
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