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ATTRv-PN: A Family Affair?

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

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Dr. Dasgupta:
This is CME on ReachMD, and I'm Dr. Noel Dasgupta. Today, I have the pleasure of speaking to Dr. John Berk about hereditary transthyretin amyloidosis with polyneuropathy.

Dr. Berk, would you mind giving us a case of a patient with transthyretin amyloidosis with polyneuropathy and discuss how you diagnosed the patient, treated the patient, and any impact on family members?

Dr. Berk:
Noel, it's always a pleasure to engage with you. Let's see what we can do here. We recently saw a 74-year-old father and a 48-year-old daughter. The father initially presented 5 years ago with long-standing carpal tunnel syndrome, lower leg paresthesias, early foot drop, regular diarrhea, and blackouts, signs of soft tissue disease, sensorimotor and autonomic neuropathies, and maybe some early cardiomyopathy. His daughter described intermittent hand numbness and subtle toe tingling when lying in bed, manifestations of carpal tunnel syndrome, and early length-dependent sensory neuropathy. Her father's diagnosis prompted the daughter to get genetic testing, confirming T60A TTR mutation. The family experience is common. Diagnosis of the index case appears to materialize out of the blue, completely unsuspected despite being a hereditary disease. Consequently, the father's disease is predictably advanced at diagnosis. The daughter's diagnosis, in contrast, is early in the course of TTR disease, reflecting anticipation of disease onset in later generations.

How do you treat family members who have different degrees of disease severity?

From my perspective, an individual's disease burden at diagnosis dictates treatment options. Early disease is often well managed by oral TTR protein stabilizers. The pill regimen is easy, but patients need close follow-up for signs of disease progression, as TTR protein stabilizers slow but rarely stop disease. TTR gene silencers, in contrast, more reliably halt TTR disease. The more severe the disease at the presentation, the more imperative it is to stop disease progression, aiming to preserve functional status.

So in conclusion, I would say when people encounter early disease, think TTR protein stabilizers. Late life-altering disease, think TTR gene silencers. And as these cases hopefully illustrate pursuing assessment and genetic testing of offspring has real value in identifying patients with early disease. If we've learned nothing else, treatment of TTR disease is most impactful when started early in the course of disease.

Dr. Dasgupta:
That was great, Dr. Berk. Thank you. For me, I learned the importance of genetic testing and identifying family members that may be at risk. It seems, from what you've told us, that treatment can be individualized based on the severity of disease and is most effective if started early.

The familial component of hereditary transthyretin amyloidosis is sometimes missed, even when patients have a family history of cardiomyopathy or neuropathy. So it's really important to consider the family history and all individuals impacted potentially, and then start trying to do therapy from there.

Well, this has been a great discussion. Thank you so much, Dr. Berk. Unfortunately, we don't have time to talk more today, but look forward to talking to you later.

Announcer:
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