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Challenges in Recognizing and Diagnosing Narcolepsy

Announcer:
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Dr. Trice:
Hi, I'm Dr. Kevin Trice with Norton Healthcare, here to talk to you today about Challenges in Recognizing and Diagnosing Narcolepsy.

Narcolepsy is a rare disorder. It affects around 0.02% of people worldwide. These are mostly adults. However, the estimated prevalence in the United States is about 1 in 2,000 patients. And this is what classifies it as a rare disease. We know that the syndrome has certain characteristics, like weight gain which can be associated with it, and that it can last a lifetime. It's not something that is curable but can be treated. Adolescents and young adults are more likely to experience this disorder in their childhood, again, but it can be present in adults later in life as well.

Narcolepsy has five major symptoms and things that we use to help characterize it. The most common one is pathologic daytime sleepiness or excessive daytime sleepiness. A second one, which can help define narcolepsy type 1 from narcolepsy type 2 is cataplexy, or the sudden loss of muscle tone associated with emotion. For instance, some people when laughing or having sadness or even joy or anger will experience muscle weakness in their arms or legs. Maybe in their mouth, they may have a drool or one side of their face may change, and this can be diagnostic of cataplexy in type 1 narcolepsy. Other symptoms would include sleep paralysis, and we'll talk more about that in just a second. A fourth one is hypnagogic or hypnopompic hallucinations. And these are visual hallucinations that tend to occur when patients are falling asleep, and sometimes when they're first arising from sleep. And the last one is disrupted nighttime sleep. Although this is a disorder of excessive daytime sleepiness, frequent nighttime arousals can also plague the patients with narcolepsy.

In terms of the sleep paralysis, this is identified as this brief loss of muscle tone, and it can occur in 75% of the time with visual hallucinations. Patients may feel like they have chest pressure, they may feel tachycardia or palpitations, they may have an out-of-body experience, and they may have this kind of intruder syndrome where they fear someone coming through the house and they just can't get their eyes open, but once they do, they know that this was a hallucination and it has passed. But the tachycardia and chest pressure may persist for several seconds to minutes after the fact.

Some of the features that happen in children can be a little bit different than they are in adults. At the onset of disease, there are a wide range of motor problems. Children can have perioral movements around their mouth, they can have dyskinetic or dystonic movements, maybe feel weak or clumsy, or even have kind of winding movements. They may have cataplexic facies; in other words, the face may kind of freeze up when they're really emotional. They may have problems in school with academic deterioration. They may have problems with memory or concentration. And they may have a lot of restlessness or motor activity, which can be one of the dominant symptoms in that group of patients.

Similar to the five symptoms, there's five different ways to do testing in patients. One of the more common ways is just to look at our

physical exam and history, followed by some objective things like the Epworth Sleepiness Scale score, overnight polysomnography in a sleep lab, followed by the Multiple Sleep Latency Test, or MSLT. This is a complex test which really looks for abnormal REM happening during sleep, particularly during the naps the second day. And then the last way to do this is a little bit more invasive; this can be done with oral swabs, sometimes bloodwork, or even a lumbar puncture, looking for certain things in the blood. These things aren't commonly done but can be very useful to help diagnose those in different cases.

So, in general, if you have a patient who has cardinal symptoms of excessive daytime sleepiness, maybe some disrupted nighttime sleep, they're not responding to typical medications, or you're having difficulty trying to understand what the etiology is, remember, although narcolepsy is a rare disorder, there are some simple features that can be common in other disorders that might lead you to further testing, as listed on this slide.

Thanks again for joining us today. I hope this was helpful.

Announcer:
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