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Early Diagnosis: Its Pivotal Role in Optimizing Outcomes in Pediatric Patients with Narcolepsy

Announcer:
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Dr. Maski:
This is CME on ReachMD. And I'm Dr. Kiran Maski. I will discuss the role of early diagnosis and optimizing outcomes in pediatric patients with narcolepsy.

It's been well known that the onset of narcolepsy typically begins in childhood with this nice histogram showing that many patients oftentimes express having symptoms before age 18. However, the diagnosis which is in black here oftentimes is not until adulthood. This decoupling of when symptoms actually begin and when symptoms are actually diagnosed is what we could call the delayed diagnosis problem. We followed this up with a more recent study to look at if delayed diagnosis is still occurring.

There are many reasons for delayed diagnosis or even misdiagnosis of narcolepsy. Patients with narcolepsy had reported that they were four times more likely to receive a diagnosis of something else rather than narcolepsy at the beginning of their journey. These included mental health problems such as depression or anxiety, or even mood or psychiatric disorders, such as, schizophrenia, epilepsy, perhaps cataplexy is mistaken for epilepsy and insomnia or other sleep disorders because of the disrupted nighttime sleep.

There was also a study showing a general lack of awareness by primary care providers, who many could not identify primary narcolepsy symptoms. And certainly it is confusing because many of the symptoms of narcolepsy can overlap with more common conditions. Also, in the background, excessive daytime sleepiness is becoming more common. In a survey of nearly 6,500 teenagers, 41% reported that they were struggling with excessive daytime sleepiness, and nearly 12% reported hypersomnolence is defined as unrefreshing, sleep, long sleep durations, and still daytime sleepiness in the morning. This makes it difficult to identify the rare condition of narcolepsy. within this background in children in specific, sometimes sleepiness might manifest as more behavioral problems,

Such attention issues, memory issues, hyperactivity, impulsivity problems, or general moodiness or emotional mobility. The core symptoms of narcolepsy, nearly all patients will report excessive daytime sleepiness, but the excessive daytime sleepiness usually presents in passive situations when kids are more sedentary. And this sometimes makes it difficult to observe the sleepiness as parents, for instance, might not be with their kids during such settings. usually if kids are interested or engaged or active, the sleepiness is less notable. Some patients do report having sleep attacks, where they are overcome with bouts of sleepiness, and those bouts might occur for anywhere from 15 to 30 minutes or be much shorter.

Hypnogogic and hypnopompic hallucinations are like the dream-like imagery that's intruding into wake states. And this is, essentially symptoms of, feeling like someone's in the room with them, a visual image of someone, a shadowy figure.

animal shapes, cartoon bubbles are also reported by kids and sometimes can be quite threatening. And sleep paralysis is this feeling where you can't move your body for a few minutes. And it's usually the paralysis is, is the rem atonia intruding during wake periods, and

it feels like a pressure like sensation, or kids sometimes report it like an elephant sitting on them making it hard to breathe. Typical cataplexy occurs in narcolepsy type one and is presented as, bilateral loss of muscle tone, usually affecting the face, neck, legs, and arms. there can be usually, it's associated with a positive emotion such as laughter, but certainly other emotions can trigger cataplexy as well as including anger, surprise, anticipation or even sometimes stress. These episodes usually last up to one minute, but are typically very brief 20, 30 seconds at most, and patients are usually fine thereafter.

They have variable frequency and that might be related to the emotional trigger content. And again, consciousness is preserved and there's abrupt return of the muscle tone after the attack.importantly, these events are fairly stereotyped for individuals and there can be partial cataplexy where it just affects the face or full cataplexy where it affects the full body and the knees and the full cataplexy can result in near collapse. So reports of what we hear about, oftentimes are when he laughs his head, bobs up and down uncontrollably. When I get upset, my knees buckle and I fall. Or there might be even the observation that the face just looks more droopy, and it looks almost drunk, like as, as terms I've heard before. So here's a cartoon of cataplexy. So here this child is experiencing laughter as a strong emotion.

Then this is an image of partial cataplexy. He has some weakness in the neck. You can see his eyes are closed, and his face sort of has a droopy like appearance. If this evolves into full cataplexy, it can affect the back muscles and the knees, and there's sort of this slow slump to the ground, because the, it's sort of a relatively slow, melting phenomenon that patients experience, they usually can grab onto something or sit down. So that injury is prevented however injury can occur. And there are risks with swimming uniquely in children. There's a phenomenon called cataplexy, facies, and this is where the cataplexy is chronic or static. It's not all, it's not triggered by emotion at all. This is just how these kids look. And this usually occurs at the beginning of the disease onset. And so you can see here this kid's presenting with bilateral posis, his mouth is hanging open, the tongue is protruding, and sometimes kids even can have these positive motor movements that look like dyskinesia or tongue.

The bribing movements or darting movements or chewing movements, again, unique to children, it's been more recently identified that there's a number of medical comorbidities of narcolepsy, specifically in children obesity. and rapid onset obesity has been reported, within months from onset of the excessive daytime sleepiness. And that occurs in 25 to 60% of patients.

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