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Respiratory Challenges in Neuromuscular Disorders: A Look at Symptoms and Impacts

### Announcer:

You're listening to *NeuroFrontiers* on ReachMD, and this episode is supported by UCB. Here's your host, Dr. Charles Turck.

### Dr. Turck:

Welcome to *NeuroFrontiers* on ReachMD. I'm Dr. Charles Turck, and joining me to examine the relationship between neuromuscular disorders and respiratory complications is Dr. Jason Ackrivo. Not only is he the Associate Director of the Fishman Program for Home Assisted Ventilation, but he's also an Assistant Professor of Medicine in Pulmonary, Allergy, and Critical Care and Neurology at the Hospital of the University of Pennsylvania. Dr. Ackrivo, thanks for being here today.

### Dr. Ackrivo:

Thanks so much for inviting me.

### Dr. Turck:

Well, to start us off, how common are respiratory complications in patients with neuromuscular disorders?

### Dr. Ackrivo:

I think it's safe to assume that it's fairly common, although clinicians should be aware that there's a long list of different types of neuromuscular disorders, from inflammatory to congenital to acquired, and so they can affect the respiratory system at varying rates. However, it would behoove any clinician evaluating somebody with any type of neuromuscular disorder to suspect that their respiratory system could be involved, and a careful history is very important.

### Dr. Turck:

So with all that in mind, would you tell us about the respiratory symptoms we would want to be on the lookout for in patients with neuromuscular disorders?

### Dr. Ackrivo:

So in the timeline of how neuromuscular disorders can affect breathing, the earliest manifestations would be overnight while the person is sleeping. It's only later on as the disease progresses that daytime symptoms can develop. So it's very important to recognize that the earliest manifestations actually affect the person's sleep. So you want to do a very careful history of the person's energy levels and whether there's any symptoms or signs that they have disturbed sleep. This could be unrefreshing sleep, waking up feeling very tired, waking up with headaches, needing to take a nap just to get through their day, and daytime fatigue. Their family members or caregivers may report that they have some confusion, or the patient feels like they just can't concentrate. Beyond that, the respiratory muscles can affect their breathing in different positions. So not only could they have shortness of breath just sitting in a chair or with minor activities like transferring from a chair to a toilet or bed, but they may have shortness of breath when lying flat. That's also called orthopnea. And so the position can highly affect their ability to feel like they're breathing okay.

If it gets really bad, some of the later manifestations will cause an elevation in the person's carbon dioxide levels, which often aren't checked, so that can secondarily cause a low oxygen level. So low oxygen, or hypoxemia, can be a warning sign. Beyond just the breathing muscles, just remember that beyond shortness of breath, the breathing muscles also affect the person's ability to clear their airways. So airway clearance, cough, and respiratory infections can be symptoms or signs that their respiratory impairment is weakened. And swallowing as well—if they're not able to swallow adequately or safely, then things can go down the wrong pipe into the lungs and cause respiratory infections.

**Dr. Turck:**

Now, continuing to look at the impact of those symptoms, how do they affect a patient's prognosis?

**Dr. Ackrivo:**

So there's a strong body of literature to show that in patients with neuromuscular disorders, particularly ALS, there's a strong link between respiratory function and the person's survival. So some of the tests that we use, such as spirometry or forced vital capacity, have correlated with survival in neuromuscular disease. We've also had a body of literature that shows that use of treatments such as noninvasive ventilation does impact the person's survival. So in ALS, for example, there was a randomized controlled trial done about 20 years ago that showed a survival benefit to using noninvasive ventilation in ALS. And subsequent studies, including our own research at the University of Pennsylvania, have shown that there is an improvement in survival for subjects who use noninvasive ventilation versus those who don't. And in addition, amongst those on noninvasive ventilation, patients who use it more hours of the day, such as more than 4 hours during a 24-hour period, have an improvement in their survival.

We also have some research that we published last year from the University of Michigan that showed that subjects who had initial elevation in their carbon dioxide level, or chronic hypercapnia, had an improvement in their survival if, with therapy, their carbon dioxide levels were able to be lowered over time.

**Dr. Turck:**

Now, you started to touch on this a little bit before, but could you paint a picture for us of how respiratory complications can affect a patient's overall quality of life?

**Dr. Ackrivo:**

One of the earliest symptoms and signs is the impairment in the quality of sleep, which ends up causing a long list of issues for patients during the daytime—they have trouble concentrating, they have constant fatigue, and they can have impaired executive function. So this can cause issues with their ability to keep appointments, pay their bills, and perform well at their jobs. It can cause an increased burden on their caregivers. They may have an increased need to seek medical help, and in the worst-case scenarios, they may end up in the hospital with respiratory complications, such as pneumonia. The patients, because of their symptoms, may hesitate to leave their room or even leave their house. However, with proper treatment, we can help them expand their world and what they're capable of doing during the day so that they can do things like go to their jobs or not be tethered to an oxygen machine. And that allows them to be more mobile and accomplish more during their day.

**Dr. Turck:**

For those just tuning in, you're listening to *NeuroFrontiers* on ReachMD. I'm Dr. Charles Turck, and I'm speaking with Dr. Jason Ackrivo about the symptoms and impacts of respiratory complications in neuromuscular disorders.

So given those impacts, Dr. Ackrivo, how can we proactively detect and monitor respiratory decline in patients with neuromuscular disorders?

**Dr. Ackrivo:**

So given that the earliest manifestations of neuromuscular respiratory impairment occur with sleep, I would start with a high index of suspicion that this person could have some symptoms or signs of impaired sleeping. That may include considering referring them for a sleep study if they have any symptoms that cause concern for the clinician. Now, you may have an issue with having a sleep center be familiar with caring for someone with neuromuscular disease, so I would always ask the sleep center how familiar they are with performing sleep studies on individuals with neuromuscular disease. And remember that patients may not volunteer that they have shortness of breath during the day as they assume that this is just part of the disease. So going along with the high index of suspicion, you have to take a careful respiratory history about whether or not they experience any shortness of breath during normal daytime activity.

In addition, there's standard-of-care respiratory testing, which includes a spirometry test in both the upright and supine position, and performing a maximum inspiratory pressure, or MIP, test that can also be in the upright and supine position. For patients who have equivocal or borderline daytime testing with spirometry or MIP values, you can request an overnight pulse oximetry, which will monitor their oxygen overnight and may be able to detect whether they are hypoventilating. Another test, which is a bit more invasive and not done as commonly, is an arterial blood gas, which will give you a direct measurement of the person's carbon dioxide level. If you have a suspicion of diaphragm function, you can refer the person for what's known as a sniff test where, under fluoroscopy, they take an inhale, and it directly shows how the diaphragm moves, which may be a sensitive test for identifying early diaphragm dysfunction. And in our center, we've also begun using transcutaneous carbon dioxide monitoring, both in the clinic and in the sleep lab, and we're doing some research now on the utility of doing that in the home as well.

**Dr. Turck:**

And before we close, Dr. Ackrivo, do you have any final thoughts on the relationship between neuromuscular disorders and respiratory function that you'd like to share?

**Dr. Ackrivo:**

You definitely want to remember that the earliest manifestations occur overnight while the person's sleeping, and if you wait until they have daytime symptoms and impairment during the day, you're already well behind the disease process affecting the respiratory function. And then beyond just ventilation and shortness of breath, remember that the weakness of the respiratory muscles can affect their airway clearance. So ask about history of pneumonias, how strong their cough is, and their ability to clear their chest, and it can also affect their upper airway and their swallowing function. So doing a good history about how well they're able to chew and swallow their food safely can help you risk stratify them for whether they may be at risk for pneumonia.

Also remember that the gene therapy landscape for these neuromuscular disorders is evolving very rapidly, and that's going to lead to more patients who are in the pediatric medicine world surviving into adulthood. So adult medicine clinicians should be aware that, in the coming decades, neuromuscular disorders are going to become more common, and therefore it's of the utmost importance that we recognize how to identify and treat respiratory complications of neuromuscular disorders.

**Dr. Turck:**

Well, as final comments bring us to the end of today's program, I want to thank my guest, Dr. Jason Ackrivo, for joining me to discuss respiratory complications in neuromuscular disorders. Dr. Ackrivo, it was great having you on the program.

**Dr. Ackrivo:**

Thank you so much for inviting me.

**Announcer:**

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