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Respiratory Dysfunction in Neuromuscular Disorders: Evaluating Symptoms

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

Dr. Turck:
This is *NeuroFrontiers* on ReachMD, and I'm Dr. Charles Turck. Joining me to discuss respiratory complications in neuromuscular disorders is Dr. Hank Mayer. Not only is he the Medical Director of the Pulmonary Function Laboratory at the Children's Hospital of Philadelphia, but he's also a Professor of Clinical Pediatrics at the University of Pennsylvania's Perelman School of Medicine. Dr. Mayer, welcome to the program.

Dr. Mayer:
Thank you very much.

Dr. Turck:
So if we start with some background, Dr. Mayer, would you tell us about the most common respiratory symptoms you encounter in patients with neuromuscular disorders?

Dr. Mayer:
There are two general categories of respiratory symptoms. The first is related to inadequate airway clearance, meaning the inability to take a deep breath and then forcefully exhale or cough. And that typically occurs as the first respiratory sign of dysfunction and will typically cause patients to require a much longer time to get over an acute illness, typically going from three to seven days to as many as two to three weeks for a common respiratory illness for them to get through it and to recover.

The second is related to the primary function of the lung, which is gas exchange, bringing in oxygen, and removing carbon dioxide. And that typically occurs after trouble with airway clearance and presents at night during deep REM sleep and then can progress through the entire duration of sleep. And that can cause patients to have inadequate sleep and then wake up feeling that they're not well rested, and that can have broader impact in terms of patients recovering from their exertion during the day and then being able to function properly. And that can be a problem if they're not able to sleep effectively at night.

Dr. Turck:
And as a follow up to that, how do you approach the initial respiratory assessment of a patient with a suspected neuromuscular disease or mitochondrial myopathy?

Dr. Mayer:
Sure. The general approach that we take is to look at everything, and it starts from watching the patient's pattern of breathing from across the room and then, with their shirt removed, looking for synchrony between the expansion of the chest and the expansion of the abdomen.

Oftentimes in patients with neuromuscular disease, as they begin to show signs of weakness, they'll have to work harder to breathe using accessory muscles. And we can see the chest going out while the abdomen's going in, and vice versa, which would be an indication of the muscle contraction in one portion of the respiratory system—like the abdomen—going out before the patient's able to generate enough force to expand the lungs.

We then look at auscultation or use our stethoscope to look at regional aeration. How well is the patient able to bring air into the lower

lobes versus the upper lobes and the right lung versus the left lung? And the other thing that is also very important is looking at respiratory rate relative to the age-appropriate norms. And if you have inefficient respiration, what happens is that you compensate by breathing more rapidly, typically in what we call a rapid shallow breathing pattern. And so if we see tachypnea—rapid breathing—then that usually makes us concerned about a patient having some type of respiratory muscle dysfunction. And when we see that, then we dig deeper to try and figure out what is going on and how to help support the patient.

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. Hank Mayer about respiratory symptoms in patients with neuromuscular disorders.

So, Dr. Meyer, let's discuss potential impacts on patients. How does the progression of respiratory decline typically differ between various neuromuscular disorders and mitochondrial myopathies?

Dr. Mayer:
I'll answer that in two ways. If you take a generic, progressive neuromuscular condition, you go through the same types of phases of respiratory dysfunction, from having trouble with airway clearance to having trouble with breathing at night to having trouble with breathing during the day. The difference among the wide range of different neuromuscular conditions is when that happens time-wise and how rapidly the different phases occur from the onset of the weakness or the condition. In other words, there can be a great difference in how rapidly progressive a condition can be. And even within a particular condition, you can have patients that have a less severe phenotype where there's very slow progression versus a more severe phenotype where there's a very rapid progression. And so, as a pulmonologist, what I focus on is looking for the different phases of respiratory dysfunction, airway clearance, and then different ventilatory or respiratory requirements for nighttime and then daytime support.

Dr. Turck:
And if we look at one more component here, Dr. Mayer, what can a pattern of muscle involvement or diaphragmatic weakness tell us?

Dr. Mayer:
Typically, diaphragm weakness can tell us a number of things. One, the patient's going to be unable to breathe in deeply. And when that happens, that can limit their ability to cough and clear secretions as efficiently as they could with better diaphragm strength. And then it also can cause them to eventually be unable to bring in enough oxygen to satisfy the body's needs and then to remove their carbon dioxide or ventilation. When that occurs, we often will see differences in breathing pattern, with more forceful chest wall motion and less abdominal motion because of the inadequate downward motion or contraction of the diaphragm.

Dr. Turck:
Well, as we approach the end of our program, Dr. Mayer, would you discuss the importance of early intervention in respiratory care for patients with progressive neuromuscular conditions or mitochondrial myopathies, such as thymidine kinase 2 deficiency?

Dr. Mayer:
Being proactive and intervening early is absolutely critical and it's a very important part of any and all standard-of-care documents for different types of neuromuscular conditions. And what that means is that as soon as a patient is showing signs of respiratory difficulty from, say, an acute illness, their caregivers need to be able to implement the respiratory support that they need. And the prime example of that would be for somebody that has inadequate airway clearance and mechanical in-exsufflation to support airway clearance.

When patients without neuromuscular disease get sick, we know that they're sick in part because they're coughing. If you are unable to cough effectively, then what can happen is that your respiratory infection can progress much further than it needs to. And in a more proactive situation, if you're able to introduce airway clearance using mechanical in-exsufflation early in the course of an illness, that can prevent the illness from progressing to the point where it requires multiple weeks of therapy to recover or needing the patient to be treated as an inpatient.

And then as far as ventilatory support, when somebody is on nighttime mechanical ventilation, begins to get sick, and develops more fatigue, it's even more critical that they have that support not only at night, but occasionally during the day. And I'll often counsel our families to have their children on daytime ventilation as a supplement to their nighttime support when they're getting sick.

Dr. Turck:
Well, given the importance of our discussion today, I want to thank my guest, Dr. Hank Mayer, for joining me for an in-depth look at some of the major respiratory symptoms associated with neuromuscular disorders. Dr. Mayer, it was great having you on the program.

Dr. Mayer:
Thank you very much. It was a pleasure.

Announcer:
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