



## Managing Respiratory Function in Neuromuscular Disorders

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Respiratory function is a critical focus for many with neuromuscular disorders. Jeanette Brown, MD, PhD, an associate professor of medicine in the department of internal medicine in the division of pulmonary and critical care at the University of Utah, takes us through critical concepts.

Neuromuscular disorders can affect respiratory function for several different reasons, including weakness of respiratory muscles, hypotonia of bulbar muscles, anatomical abnormalities (e.g., scoliosis or rigid spine), and decreased central respiratory drive.

Amyotrophic lateral sclerosis (ALS) is the most common neuromuscular disease that significantly impacts respiratory function, but spinal muscle atrophy (SMA), Duchenne muscular dystrophy (DMD) and many others can also cause respiratory issues that require monitoring and management.

“Depending on the type of underlying disease, the kind of impact it will have will be based on the age of the patient when symptoms first presented and the location of symptom onset,” Dr. Brown says.

## Starting Respiratory Management

When to begin respiratory monitoring and management should be decided based on the disease type and on a case-by-case basis. “People are on a spectrum, but typically once a patient can’t ambulate it’s time to start worrying about needing respiratory support,” Dr. Brown says.

In the meantime, “We do routine screening with lung function tests and try to pick these issues up before a patient is symptomatic,” she says.

## Preserving Respiratory Function

It’s important to begin preventative measures to help preserve respiratory function as early as possible. For example, for children with SMA, gene therapy administered early can preserve muscle strength and respiratory function before they are lost.

“With newborn screening, you can find issues before the patient is symptomatic and through care can develop neurological milestones,” Dr. Brown says. SMA and Pompe disease are the only neuromuscular diseases currently included in the Recommended Uniform Screening Panel (RUSP), but MDA is advocating for adding DMD and other neuromuscular diseases for which effective therapies exist.

“For adult patients with ALS or SMA, treatment is more about trying to preserve function and slow decline,” Dr. Brown says.

## Managing Respiratory Issues

The decline of respiratory function can be very slow, depending on the type of neuromuscular disease. However, it’s not unusual for a health issue to hasten that decline. “It can be many things that accelerate the need for mechanical intervention, including having a weak cough and contracting COVID-19, influenza, or parainfluenza viruses,” Dr. Brown says.

In the case where an infection leads to exhaustion from coughing and struggling to breathe, mechanical insufflation-exsufflation aids (brand names include CoughAssist, VitalCough, and BiWaze Cough System) and other devices are used to mechanically assist the patient to cough, re-expanding the lungs and pulling secretions from the bottom to the point where they can get them out.

Some patients benefit from using non-invasive positive pressure ventilation, such as bilevel

positive airway pressure (BiPAP), delivered using a mask. They may start using it at night and progress to using it in the daytime as well.

When a patient needs respiratory management, it's important to include a respiratory therapist and pulmonologist (pediatric or adult) on their care team. These providers can monitor lung function over time and provide education on daily and emergency care and managing symptoms when a patient is sick. Providers can also provide training on using devices at home.

However, more important than who is on the team is that all the providers work together. "Overall, we are all a part of the same team, trying to make quality of life the best it can be for our patients and caregivers," Dr. Brown says.

