

# Breathing Problems in Children with Neuromuscular Diseases

## Part I

Neuromuscular diseases are diseases of the nervous system (nerves that control the body) and muscles. Neuromuscular diseases (as a group) are common, with about 1 in 3000 children being affected. These conditions range from mild, causing few problems to the child's daily life, to progressive and severe, making it difficult to walk or move around without help, swallow well, or sleep without help from a device. Examples of neuromuscular diseases are: *spinal muscular atrophy (also called Werdnig-Hoffman disease)*, *Duchenne (doo-shen) muscular dystrophy*, *congenital muscular dystrophy* and *congenital myopathies*. Most of these disorders are genetic or inherited (passed down in a family). Most are first noticed during early childhood but are not always obvious at birth.



Neuromuscular conditions can cause breathing problems in children in several ways—directly and indirectly (because of related complications). This fact sheet describes how neuromuscular problems can cause breathing problems in children. For more on how to treat these breathing problems, see part 2 at [www.thoracic.org/patients](http://www.thoracic.org/patients).

### What muscles are used in breathing?

A number of different muscle groups are important to breathing. These include diaphragms and muscles of the chest, back, and upper airway. The chest muscles and diaphragm are important for breathing and coughing. The back muscles keep the spine straight. The upper airway includes the nose, mouth, tongue, back of the throat (pharynx), voice box (larynx) and epiglottis. The epiglottis is the flap that keeps food from going into the windpipe with swallowing. Muscle weakness and poor muscle coordination can affect any part of the upper airway and cause breathing problems.

### How do neuromuscular diseases cause breathing problems?

Neuromuscular disorders can cause breathing problems in one or more of the following ways. These problems may appear at different ages and progress over time.

1. *Reduced lung function and hypoventilation.* With hypoventilation, the amount of air going in and out of the lungs is reduced. This can lead to lower oxygen levels

and higher carbon dioxide levels in the body. Weakness of the diaphragms and other breathing muscles restricts the chest movement and reduces lung function. If these muscles don't work well, your child cannot take as deep a breath as usual. Scoliosis (curve of the spine) can develop with muscle weakness and limit the chest size and ability to take a deep breath.

2. *Weak cough and difficulty clearing mucus from the airways.* Muscle weakness can make it difficult to get mucus (phlegm) out of the lungs leading to airway mucus plugging and chest congestion. Coughing helps keep the airways clear of mucus. A strong effective cough requires the diaphragms, chest and throat muscles to work together. Mucus that stays in the lungs can block the airways and cause atelectasis (collapse of air sacs in parts of the lungs) and make it hard to get enough oxygen. It is also easier to get a lung infection. Some children only have mucus problems when they get a cold or infection, when a good cough is needed the most. The child may feel the need to cough but not have enough strength to do it well.
3. *Swallowing problems and risk of choking.* Muscle weakness can affect how a child swallows and keeps fluid or food from going down the windpipe. If fluid, food or saliva go into the windpipe, this is called aspiration. Aspiration can cause inflammation (irritation) of the airways, infection, and scarring of the lungs. Swallowing

problems can vary from choking only with thin liquids to being unable to even clear saliva from the throat. Some children have swallowing risk only at times when they are sick and weaker than usual. Unfortunately, some children can have 'silent' aspiration in which they do not have a protective cough when the food or fluid goes into the airway, but have breathing problems after it occurs.

4. **Upper airway obstruction and sleep apnea.** The airway muscles normally are more relaxed during sleep, but if there is muscle weakness, this can result in blockage of airflow and obstructive sleep apnea (OSA). OSA can cause serious health problems and poor sleep quality.

### What are symptoms of neuromuscular related breathing problems?

Symptoms can include:

- fast shallow breathing
- increased use of other muscles such as in the neck or rib cage (retractions)
- fatigue
- headaches (especially in the morning)
- weak cough
- choking

However, there can be breathing problems that are serious but do not cause immediate symptoms. A child may not complain about feeling short of breath or work hard to breathe even with low lung function.

### What tests are done to look for breathing related problems in neuromuscular disease?

Chest x-rays can help show the size of the child's chest and whether there is any mucus plugging in the lungs. If the chest wall muscles don't work well, the chest appears smaller on x-ray. Pneumonia and lung changes from aspiration can be seen on x-ray. If there are no changes seen on an x-ray, it is still possible to have a breathing problem.

Breathing function tests, called pulmonary function tests, can be done in children, usually 6 years and older. These include measures of respiratory muscle strength and spirometry. Your healthcare provider may do these breathing tests every year or more often to help measure how much reserve and lung function your child has while awake.

Your child's oxygen level (*oxygen saturation* or "*O<sub>2</sub> Sat*") can be measured with a small device called a *pulse oximeter*. This device can be clipped painlessly on to your child's finger, toe or earlobe. Oxygen saturation can be checked at various times such as during sleep or with exercise.

Sleep studies are a way to assess your child's breathing during sleep. The carbon dioxide level (CO<sub>2</sub>) can be measured in the blood. If you do not breathe well (hypoventilate) your carbon dioxide level will be high and can cause acid balance problems in the body. A blood gas is done to measure the CO<sub>2</sub> level.

Upper airway muscle function and swallowing ability can be assessed with a swallow function study, which is an x-ray imaging study done together with feeding observations by a speech pathologist. Sometimes a procedure (laryngoscopy) is done to look at the upper airway and vocal cords.

Scoliosis is screened for on physical exam and with spine x-rays. Your child's healthcare provider will decide when x-rays are needed and if your child needs to see an orthopedic surgeon for scoliosis.

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## Rx Action Steps

- ✓ If your child has a neuromuscular disease, talk with your healthcare provider about symptoms to watch and how often tests should be done to check your child for breathing problems.
- ✓ Work with your child's healthcare provider to address conditions that can affect breathing such as swallowing problems or scoliosis.
- ✓ Wash your hands often and well when caring for your child. Use soap and water or alcohol-based hand sanitizer.
- ✓ Avoid having your child around people who are sick. Keep your child away from tobacco smoke.
- ✓ Have your child and caregivers get a yearly flu vaccine.

**Healthcare Provider's Contact Number:**

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## For More Information

### American Thoracic Society

- [www.thoracic.org/patients](http://www.thoracic.org/patients)
  - Lung function tests in Children
  - Obstructive Sleep Apnea in Children
  - Pulse oximetry
  - Sleep studies in Children

### Muscular Dystrophy Association

- <http://mda.org/disease>

### The Cooperative International Neuromuscular Research Group

- <https://cinrgresearch.org/>

### National Library of Medicine Medline Plus

- <http://www.nlm.nih.gov/medlineplus/neuromusculardisorders.html>

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