

Breathing Problems in Children with Neuromuscular Diseases

Mini-series 1 of 2

Neuromuscular diseases are diseases of the nervous system (nerves that control the body) and muscles. Nerves also tell our muscles what to do, like walking. We have nerves that control our breathing without us having to think about it. We use a number of muscles to breathe including the diaphragms, chest, neck, and throat muscles. Neuromuscular conditions can cause breathing problems in several ways—directly and indirectly (because of related complications).



Neuromuscular diseases (as a group) are pretty common, with about 1 in 3000 children being affected. These conditions can range from being 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. Some 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.

This information sheet describes common breathing problems seen in neuromuscular disease and what is often done to test for and treat breathing problems.

How does muscle weakness affect breathing and what are the symptoms?

Neuromuscular weakness can affect different muscle groups important to breathing. The chest muscles and diaphragm are used to bring air in and out and also help with coughing. The muscles in the back help keep the spine straight, which is also important for chest size and expansion with breathing. The upper airway includes the nose, mouth, tongue, back of the throat (called the pharynx), voice box (the larynx) and epiglottis (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.

There are times when breathing problems are obvious with neuromuscular disease. 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, and 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. That is why tests for lung function and for breathing during sleep are recommended to look for problems that may not be obvious.

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 can develop with muscle weakness problems. 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. Hypoventilation can occur in several ways with neuromuscular disease. Weakness of the diaphragms and other breathing muscles leads to restricted 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 in neuromuscular disease and limit the chest size and ability to take a deep breath.
2. Muscle weakness and a weak cough can make it difficult to get mucus (phlegm) out of the lungs leading to airway mucus plugging and chest congestion. Coughing is a natural way that our body 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 when there is extra mucus for bacteria to grow in. 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 is not able to cough up mucus.
3. Swallowing problems from muscle weakness lead to a risk of choking. When food or fluid goes into

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the windpipe from the throat with eating it is called aspiration. Sometimes saliva from the mouth may be aspirated as well. This can happen when upper airway muscles that normally protect the airway (and therefore the lungs) are not working well. Aspiration can cause inflammation (irritation) of the airways, infection, and scarring of the lungs. Some children with neuromuscular disease have very poor swallowing ability. Others may have problems only with thin liquids that go too quickly down the throat. Some 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. Weakness in the upper airway muscles can lead to a blockage (obstruction) of airflow, particularly during sleep. The airway muscles normally are more relaxed during sleep, but if there is muscle weakness, this can result in obstructive sleep apnea (OSA). OSA can cause serious health problems and poor sleep quality. (For more information, see ATS Patient Series *Sleep Problems in Children with Neuromuscular Diseases and Obstructive Sleep Apnea in Children*).

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 or 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. However, the absence of changes on chest x-ray does not mean there may not be a breathing problem.

Breathing function tests can be done in children, usually 6 years and older. These include measures of respiratory muscle strength and spirometry. Your health care provider may do these breathing tests every year or more often to help measure how much reserve and lung function your child has awake. (see also ATS Patient Information Series *Pulmonary Function Tests in Children*).

The child’s oxygen and carbon dioxide levels can be measured in his or her blood. Your child’s oxygen level (*oxygen saturation* or “*O₂ 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. (see also ATS Patient Information Series *Pulse Oximetry*)

Another test that is sometimes done is called an arterial blood gas (ABG). In this test, blood is drawn from an artery (usually in the wrist). This can give the blood oxygen level and the carbon dioxide level. A capillary blood gas (CBG) from a finger prick or venous blood gas (VBG) also may be done to check the carbon dioxide 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 laryngoscopy (a flexible tube) 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 health care provider will decide when x-rays are needed and if your child needs to see an orthopedic surgeon for scoliosis.

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R_x Action Steps

- ✓ If your child has a neuromuscular disease, talk with your health care provider about symptoms to watch and how often tests should be done to check your child for breathing problems.
- ✓ Work with your child’s health care 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.

Doctor’s Office Telephone:

Other Resources

Muscular Dystrophy Association

<http://mda.org/disease>

The Cooperative International Neuromuscular Research Group

<http://www.cinrgresearch.org/aboutnd/diseases.cfm>

National Library of Medicine Medline Plus

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

Hull J, et al. British Thoracic Society guideline for respiratory management of children with neuromuscular weakness. *Thorax* 2012;67:i1 e i40. Access at <https://www.brit-thoracic.org.uk/document-library/clinical-information/children-with-nmw/children-with-nmw-guideline/respiratory-management-of-children-with-neuromuscular-weakness-guideline/>

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