Caring for the respiratory health of people living with Duchenne

The more you know about living with Duchenne muscular dystrophy (DMD), the better prepared you can be.
To establish a starting point, get an early reading of lung function while your loved one is still ambulatory.

Once he is non-ambulatory:
- Make sure he sees a pulmonologist twice a year.
- Look for respiratory symptoms of DMD, such as:
  - weak cough
  - fatigue
  - headaches
- Stay on the lookout for daytime fatigue and sleep problems, such as:
  - restless sleep
  - shallow breathing at night
  - nightmares
- Keep an eye on colds—they can turn into something more serious or last longer.

- Make sure your loved one has his flu and pneumonia vaccinations.
- Sign up for more information at www.TakeabreathDMD.com.

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Understanding DMD and respiratory health

DMD is a genetic disease that typically affects boys. It is caused by a lack of a protein called dystrophin. Dystrophin protects muscle fibers from breaking down. Over time, DMD causes a loss of muscle strength. Usually the leg muscles are affected first. As boys grow older, heart and respiratory muscles are also affected.

The right support and information helps people living with DMD manage lung function and well-being.

MANAGING RESPIRATORY HEALTH IN DMD
When the respiratory muscles weaken, complications may happen more often. Someone with DMD may not be able to cough or breathe strongly. This can lead to pneumonia and other serious breathing problems. These complications may be preventable by closely watching respiratory function and talking to a doctor as soon as they arise.

WHEN TO SEE A PULMONOLOGIST
Those with DMD should see a specialist in respiratory care (a pulmonologist):

- While still able to walk, boys should have a respiratory test
  - The care team can use this test as a baseline and compare the results to future respiratory tests
- Two times a year once he becomes non-ambulatory
- Every 3 to 6 months when using mechanical devices to help breathe or clear his airways or as recommended by a pulmonologist

THE DMD MULTIDISCIPLINARY TEAM
Being actively involved with your loved one’s care team can help you manage DMD. This includes understanding respiratory tests and how breathing devices work. Having many experts on the team allows for cross-specialty coordination and communication. The goal is to provide broader care and support.

A DMD care team usually includes:

- **Pulmonologist**: Specializes in caring for the lungs
- **Cardiologist**: Specializes in caring for the heart
- **Neurologist**: Treats neurological conditions and is central to your care team
- **Genetic Counselor**: Can help you understand how DMD is inherited and order genetic tests
- **Social Worker**: Helps families find medical resources and advocates for them
- **Nutritionist**: Expert on dietary needs
- **Physical Therapist**: Develops treatment techniques to improve range of motion and prevent contractures
- **Pediatrician/Primary Care Physician**: Manage non-DMD related health conditions

OTHER HEALTHCARE PROFESSIONALS:
- **Physical Medicine and Rehabilitation**: Physicians who help improve the muscles’ ability to move
- **Respiratory Therapist**: Assesses lung function and helps develop a treatment plan
- **Speech Language Pathologist**: Evaluates speech and swallowing skills, and develops treatment plan
In the early stages, the effects of DMD on respiratory health can be hard to notice. With time they become more noticeable.

**DIAGNOSIS**
At diagnosis most boys with DMD have normal respiratory function.

**AMBULATORY**
As long as your loved one with DMD is able to walk, he is less likely to have serious respiratory complications. Even so, focusing on respiratory health and getting an initial measure of lung function now could have benefits in the future.

**EARLY NON-AMBULATORY**
Once a boy with DMD begins using a wheelchair, the risk of respiratory issues increases. But early signs that respiratory muscles are weakening aren’t always obvious.

**LATE NON-AMBULATORY**
People living with DMD have a higher risk of facing a range of respiratory problems. On the following pages you can read about some of the symptoms they may face.

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**THE MUSCLES THAT MAKE BREATHING POSSIBLE**
It’s important to understand the role that these muscles play and what they mean for DMD.

The lungs can’t breathe on their own. They need coordinated movement of a collection of muscles. The muscles help expand and contract to move air in and out of the body.

**DIAPHRAGM**
Located at the bottom of the rib cage, the diaphragm is the most important muscle for inhaling air. When it moves down, it pulls air into the lungs.

**INTERCOSTAL MUSCLES**
Located between the ribs, these muscles help expand and contract the chest cavity. This allows air to go in and out of the lungs.

**ABDOMINAL MUSCLES**
These muscles help air leave the lungs. When they contract, they push the diaphragm upwards against the lungs, which forces air out.

As DMD progresses, some boys will develop scoliosis (sideways curvature of the spine). This changes their posture. It can also make it harder for weakened lungs to do their job properly.
Respiratory symptoms of DMD

The early signs of respiratory issues in people with DMD can be easy to miss. You may begin to notice respiratory symptoms once your loved one begins using a wheelchair.

EARLY SYMPTOMS
Respiratory muscles decline over time. The first signs may not appear to be tied to the lungs. They include:

- Headaches
- Fatigue
- Restless sleep
- Nightmares
- Difficulty concentrating
- Not feeling rested
- Difficulty staying awake

LATER SYMPTOMS
Eventually respiratory-related symptoms begin to show. These symptoms can be subtle—like not being able to cough hard enough—but may become more serious.

Ineffective cough
Think of coughing as the body’s way of clearing the airways. When mucus and other secretions build up, coughing helps remove them.

When respiratory muscles weaken, they can’t create a cough strong enough to clear the airways of mucus. This can lead to:

- common colds, which can become infections like sinusitis
- something more serious like pneumonia

Sleep-disordered breathing
Occurs when there are sleep problems because of abnormal breathing patterns. Because of their weakened respiratory muscles, someone with DMD might be more prone to sleep problems. But if you are aware of the signs, you can be on the lookout for nighttime breathing issues.

- Obstructive sleep apnea (OSA)
  OSA is usually caused by weakness of the upper airway muscles which can collapse. It can:
  - interrupt sleep
  - cause a drop in oxygen (hypoxemia)
  - elevate carbon dioxide levels (hypercapnia) in the blood
  Snoring is also a symptom of OSA, which can lead to waking up frequently during the night.

- Shallow breathing at night
  Normally when sleeping, breathing slows down. With DMD, normal slower breathing is complicated by weakened muscles. This causes even shallower breathing. Shallow breathing can cause hypoventilation, which is the inability to:
  - breathe deeply enough to bring in enough oxygen
  - remove carbon dioxide that the body makes
  This can lead to restless sleep and daytime fatigue in people with DMD.

Atelectasis [Aht-ell-lect-tuh-sis]
This occurs when a part of the lung “collapses” and doesn’t inflate properly. A partial collapse in the lung can occur when an airway becomes blocked, typically with mucus. The cough is not strong enough to clear mucus.
Monitoring respiratory health with DMD

Doctors check respiratory function because it can:

- help signal breathing problems now or in the future
- show respiratory muscle strength
- help measure disease progression

THE MOST COMMON MEASURES OF RESPIRATORY HEALTH:

- **FORCED VITAL CAPACITY (FVC)**: The total amount forcibly blown out after one big breath. High FVC scores are a sign that the lungs are inflating to capacity.
- **FORCED EXPIRATORY VOLUME (FEV)**: When a person exhales, this is the amount of air blown out in one second. High FEV scores are another way to see if the lungs are expanding the way they should.
- **PEAK EXPIRATORY FLOW (PEF)**: Measures the peak or maximum flow of air when a person breathes out as hard as he can.
- **MAXIMAL INSPIRATORY PRESSURE (MIP)**: Measures how strongly someone can breathe in.
- **MAXIMAL EXPIRATORY PRESSURE (MEP)**: Measures how strongly someone can breathe out.

TESTS TO MEASURE RESPIRATORY HEALTH

**Spirometry**
FVC, FEV1, and PEF are all measured by spirometry. It’s one of the most common tests used to learn how the respiratory muscles are working. Spirometry tests can:

- measure how DMD is progressing
- influence treatment decisions

**Overnight monitoring**
The first signs of weakening respiratory muscles can appear at night. Doing tests while the patient is asleep lets doctors see how well respiratory muscles are working.

- **Polysomnography (polly-sum-nog-rah-fee)**
The gold standard in assessing sleep-related breathing problems. This test can assess how the respiratory muscles and lungs:
  - bring in oxygen
  - remove carbon dioxide

- **Oximetry**
A test that can be performed at home. A sensor is attached to the finger or earlobe while the patient is asleep. This test measures the oxygen saturation levels to see if there is enough oxygen in the blood.
A cough involves bringing air in and forcing it out of the lungs. As respiratory muscles weaken, he may not be able to cough or breathe well. In addition, infections and complications can last longer.

A strong cough clears mucus from the lungs. This helps reduce the risk of atelectasis and pneumonia.

**A STRONG COUGH INVOLVES 3 PHASES:**

1. **Deep breath in**
2. **Forceful breath out, at first with the vocal cords closed for a second to build up pressure**
3. **The vocal cords open and there’s a large flow of air outward**

**UPPER RESPIRATORY INFECTIONS**

The upper respiratory tract includes the nose, nasal cavity, and throat.

The most frequent infection in the upper respiratory tract is the common cold. A cold can quickly turn into sinusitis or become more serious and move into the lower respiratory tract.

**LOWER RESPIRATORY INFECTIONS**

The lower respiratory tract includes the windpipe and the air sacs in the lungs.

Infections in the lower respiratory system are usually caused by a virus or bacteria. Serious lower respiratory illnesses include bronchitis and pneumonia.

**IMPORTANCE OF RESPIRATORY MONITORING**

Respiratory issues are a leading cause of death in those with DMD. There are a number of things you can do to care for your loved one’s respiratory health. Making sure he has a flu and pneumonia vaccine every year is important. Also important is to consult your loved one’s care team to create a care plan specific to his needs.
Sign up for more information

Stay up-to-date on the latest information about respiratory health in DMD.

Visit www.TakeabreathDMD.com and sign up to receive news alerts, updates about DMD, and more.