
Breathtaking
BREATHE EASY
Respiratory Care in
Neuromuscular Disorders
MDA
ESN
Metamorphosis

The Muscular Dystrophy Association is a voluntary health agency aimed at conquering more than 40 neuromuscular diseases that affect more than a million Americans. MDA combats neuromuscular diseases through programs of worldwide research, comprehensive medical and community services, advocacy, and far-reaching professional and public health education.

With national headquarters in Tucson, Ariz., MDA has more than 200 field offices across the country, maintains a nationwide network of clinics and supports more than 330 research projects around the world.

ON THE COVER:

"Breathtaking Metamorphosis" by Erin Brady Worsham of Nashville, Tenn., depicts her newfound freedom as a result of using a ventilator for breathing assistance. Worsham received a diagnosis of ALS in 1994 and underwent a tracheostomy in 1997. Worsham creates vibrant computer art, and "Breathtaking Metamorphosis" is one of her pieces in the MDA Art Collection.

Breathe Easy: Respiratory Care in Neuromuscular Disorders

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For copies of "Breathe Easy," contact any MDA office.

Breathe Easy: Respiratory Care in Neuromuscular Disorders

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DEAR FRIENDS:

The first symptoms of my neuromuscular disease appeared when I was 6 years old. I was well into adulthood when an MDA clinic physician diagnosed my condition as limb-girdle muscular dystrophy.

Knowing that I'd probably live into my 60s or longer, I pursued my education and my life, using a power wheelchair to get around. My career brought me from the University of Northern Colorado, my alma mater and first employer, to Arizona State University in Tempe, where I directed the Disability Resource Center for students with disabilities for 25 years until retirement in 2005. I worked 50 to 60 hours a week, supervising a staff of 40 who provided services for 1,800 students a year. My life was filled with challenges and rewards, and I loved it.

Then, in 1985, I came down with pneumonia. I recovered fairly quickly, but soon I began having memory problems and falling asleep in staff meetings. I was bone tired all the time. I was in my 40s, and I just thought I was getting old.

In 1988, I developed a bad cold, had difficulty breathing, and noticed that my lips and fingertips were blue. In the emergency room, a tube was put down my throat, and I was sent to the intensive care unit. The diagnosis was pneumonia, complicated by severe respiratory insufficiency from weakened diaphragm muscles.

Up to that point, I'd been very healthy (I thought) and hadn't taken time from my busy schedule to go to the MDA clinic for regular checkups. I didn't know that I was experiencing the symptoms of respiratory insufficiency.

After nearly two months in the hospital, I was released with a tracheostomy (breathing tube in the trachea) and a portable ventilator. Later, I got a speaking valve, but at first I was unable to speak and communicated with pad and pencil. Despite medical predictions, I was back at work within four months, after learning everything

I could about ventilation and related equipment.

In 1995, after extensive personal research and medical evaluation, I was able to switch from my trach to a noninvasive method of assisted ventilation. I still use an inflatable belt to help me breathe most of the time, as well as several oral interfaces for mechanical ventilator during the day and a custom nosepiece for nighttime ventilation.

Tedde Scharf uses a mouthpiece with her ventilator during the day.

Following retirement in 2005, I moved to Tubac, Ariz., a small, historic arts community forty miles south of Tucson. In Tubac, my sister and I opened an independent bookstore, TJ's Tortuga Books & Coffee Beans, in 2009. A whole new life experience has begun!

I've successfully used invasive and noninvasive ventilation methods for over 20 years. For those of us with neuromuscular diseases, both approaches to breathing support help maintain energy, improve the quality of life and — most important — give us more years to be productive citizens.

No matter how you and your doctor decide to handle your respiratory needs, knowledge will help overcome fear. This booklet tells how, with your MDA clinic staff, you can monitor your respiratory health. It provides common symptoms of respiratory distress. With this knowledge, you can avoid an unexpected crisis.

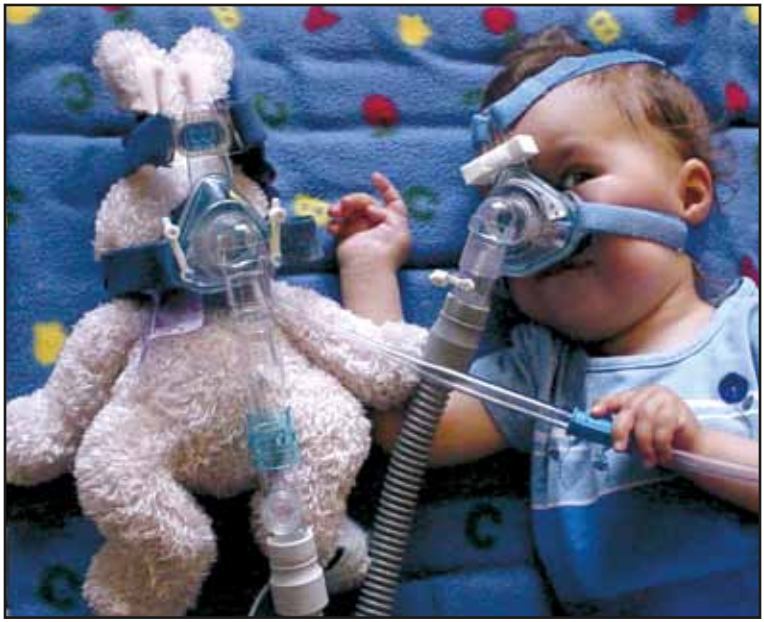
If you ever feel lost or afraid, have questions about ventilatory support or just want to share experiences, please contact me at tedde@asu.edu. If you need assisted ventilation, learn about all the options, their benefits and drawbacks.

Tedde Scharf

Tedde Sharf
Tempe, Ariz.

INTRODUCTION

Respiratory health is a vital issue for children and adults who have neuromuscular diseases. These diseases progressively weaken muscles, sometimes including those you use in breathing and coughing. Weak respiratory muscles may eventually lead to respiratory failure, a frequent cause of death in many neuromuscular disorders.



Even very young children can learn to accept respiratory care as part of life.

Fortunately, today, expert physicians such as those at MDA clinics know a great deal about treating the respiratory effects of neuromuscular diseases. This booklet will help you understand how to manage your respiratory health and intervene before it reaches a crisis level, under the direction of your health care team.

This publication is an outgrowth of MDA's "Breathe Easy: Respiratory Care for Children With Muscular Dystrophy." We're indebted to the authors of that booklet: Robert Warren, M.D.; Vikki Stefans, M.D.; and Sheila Horan, registered respiratory therapist, all at Arkansas Children's Hospital, where their work with children at MDA's clinic inspired this booklet. It proved so helpful to families of children served by MDA that we decided to expand it to cover both children and adults with neuromuscular diseases.

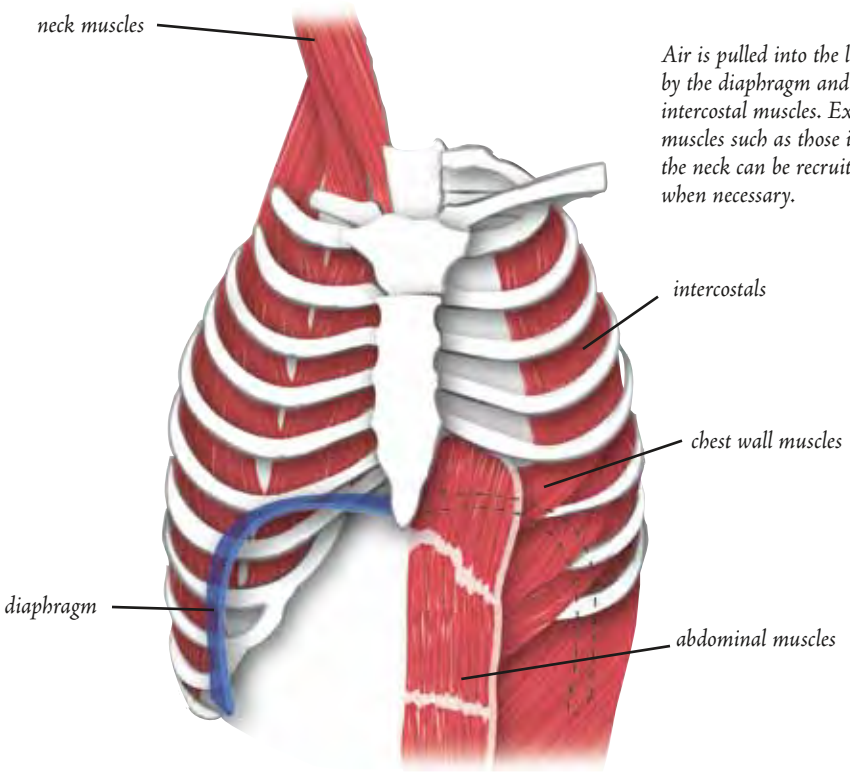
For special help with this edition, we're grateful to two expert physicians. Jonathan Finder, M.D., is a pediatric pulmonologist at Children's Hospital of Pittsburgh, where he's a consultant to the MDA clinic, and an associate professor at the University of Pittsburgh School of Medicine. Joshua Benditt, M.D., is director of Respiratory Care Services at the University of Washington Medical Center in Seattle, and sees patients at the university's MDA/ALS center and MDA clinics. He's a professor of Medicine at the University of Washington.

The contents of this booklet will raise many topics and questions that you can discuss with your MDA clinic doctor. MDA's clinics serve Americans with more than 40 neuromuscular diseases. See page 37 for other respiratory care resources.

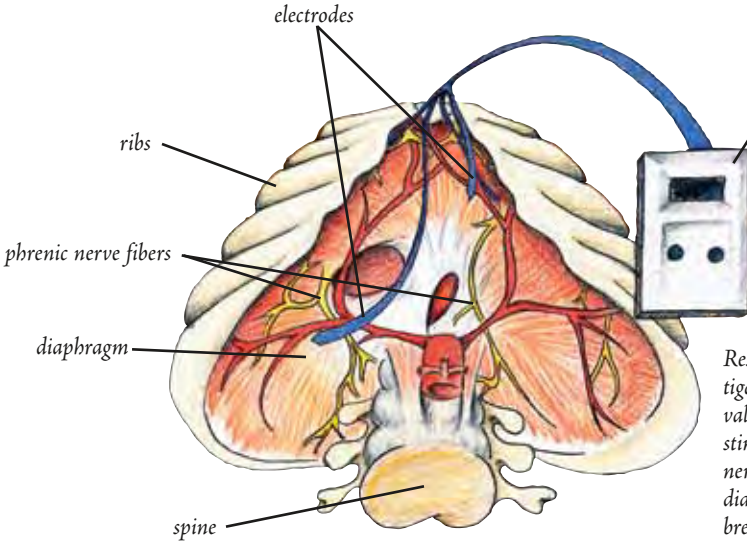
NEUROMUSCULAR DISEASES AND BREATHING

Breathing is accomplished with the effort of respiratory muscles. A dome-shaped muscle called the *diaphragm*, located below the lungs, and muscles between the ribs called the *intercostals*, pull air into the lungs. This inspiration, or inhalation, is followed by a relaxation of these muscles as air leaves the lungs.

The process of breathing in and out, taking in oxygen and exhaling carbon dioxide, is called *air (*or *gas) exchange,* or *ventilation.* The pathway that air follows from the nose, through the trachea and into the lungs is the *airway*.



Air is pulled into the lungs by the diaphragm and intercostal muscles. Extra muscles such as those in the neck can be recruited when necessary.



Researchers are investigating the possible value of electronically stimulating the phrenic nerve, which controls the diaphragm, in assisting breathing in ALS.

When you forcefully inhale or exhale, you use extra, or accessory, muscles of breathing. Accessory respiratory muscles include those in the abdomen, chest wall and neck.

Over time, neuromuscular disorders can weaken all of these muscles. When these muscles don't move, air can't move in and out of the lungs effectively.

Respiratory failure, often in association with an infection (pneumonia), is a frequent cause of death for people with neuromuscular disorders.

Sometimes a decline in respiratory function can be so gradual that it isn't noticed at all or is attributed to some other cause, such as general fatigue, depression or sleep problems.

In this booklet, you'll learn that monitoring your breathing function isn't harder than any other type of monitoring; and that with good health care, therapeutic techniques and supportive equipment, you can prolong breathing function for years and avoid having a respiratory crisis.

RESPIRATORY EVALUATIONS

Tests to evaluate breathing — known as *pulmonary function tests* — are generally noninvasive; that is, they require no needles or penetration of the body. In these kinds of tests, you breathe into a computerized machine through a mouthpiece while a clip blocks your nose.

The tests can be performed on adults or on children who are mature enough to follow instructions and cooperate with the respiratory therapist.

Many doctors recommend getting breathing tests as close to the initial diagnosis of a neuromuscular disease as possible. These tests will provide baseline measures, which can then be used to document changes and the rate of change.

A standard breathing test measures forced vital capacity, which is how much air can be expelled after a deep breath.

Your MDA clinic doctor (usually a *neurologist*) can recommend a *pulmonologist* (lung specialist) or *respiratory therapist* (a technician who performs breathing tests and treatments) who works with people who have neuromuscular diseases. They'll perform the tests you'll need to monitor your respiratory health.

Based on the results of these tests over time, your pulmonologist can plot the function of your respira-

PULMONARY FUNCTION TESTS

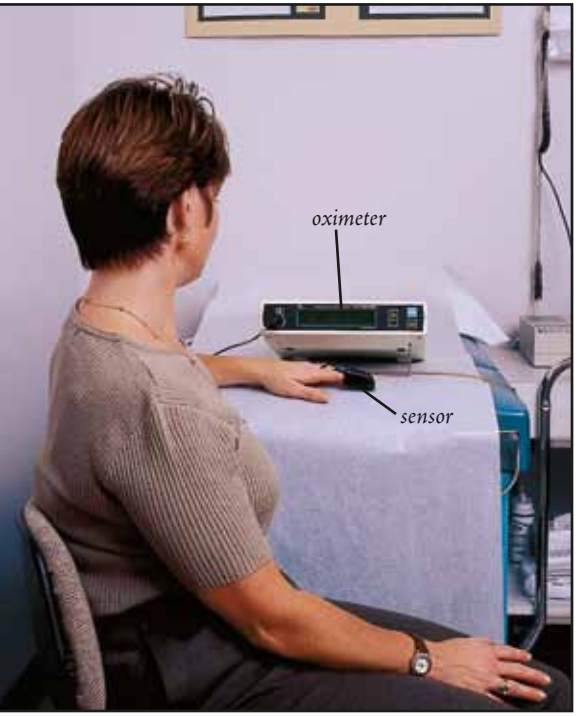
A physician may order these tests at various stages of a neuromuscular disease. Some tests require that a child be old enough to follow directions.

- forced vital capacity (FVC), which measures how much air a person can expel as fast as possible after taking a deep breath, and other capacities and volumes
- peak cough flow (measurement of how fast air is expelled as a person coughs)
- oximetry, which measures the amount of oxygen carried by the red blood cells
- overnight oximetry, which measures changes in oxygen levels during sleep
- capnography, which measures the amount of carbon dioxide in a breath
- (under some circumstances) capillary or arterial blood gases, which measure oxygen, carbon dioxide and bicarbonate levels in the blood. A finger prick or blood from a vein is needed to get blood for capillary blood gas tests.

tory system. When the numbers and physical exam show a decline, it may be time to initiate some form of ventilatory assistance to move more air into and out of the lungs (see "Respiratory Treatments," page 21).

By understanding the rate of decline in your breathing muscle function, with the help of your health care team, you can carefully consider decisions about the best form of assistance before a breathing crisis develops.

Periodic evaluations of respiratory status can assist the neurologist and pulmonary doctor in determining when to begin a particular treatment. Following a



Oximetry painlessly measures how much oxygen is in the blood through a sensor on the finger.

careful review of the physical examination and pulmonary function tests, an individual respiratory care plan can be put together for you.

In 2004, the American Thoracic Society, in consultation with experts in *Duchenne muscular dystrophy*, many of whom are associated with MDA clinics, released specific guidelines for respiratory care in this severe, childhood-onset muscular dystrophy. (See page 31.)

Guidelines for *amyotrophic lateral sclerosis (ALS)*, a paralyzing disease that destroys nerve cells in the brain and spinal cord and usually begins in late middle age, were developed by the American Academy of Neurology in 1999. They're undergoing revision as knowledge is gained. (See page 33.)

These general guidelines can serve as a starting point for doctors caring for children or adults with other neuromuscular diseases.

Some newer oximeters are very small.

PREVENTION

Can respiratory problems be prevented? Yes, but only to a certain extent and for a limited amount of time, if the neuromuscular disease attacks the respiratory muscles.

A little common sense, being prepared for emergencies with backup plans and equipment, and not taking unnecessary chances will take you a long way. For example, get a flu shot (influenza vaccine), and avoid extended exposure to people with colds. Be familiar with the symptoms of chronic underventilation (page 19) so you can speak with your doctor if they arise.

The keys to limiting respiratory problems are:

STAY WELL

Avoid colds and other respiratory infections as much as is practical.

GET IMMUNIZED

Keep routine childhood immunizations (such as those against polio, measles and mumps) up to date, with advice from your doctor.

Get a yearly flu shot if you're a child or adult

Flu shots are important for people with neuromuscular disorders, especially if they have weakened respiratory muscles.

with a neuromuscular disease, or a family member. Ask about pneumonia vaccinations.

COUGH

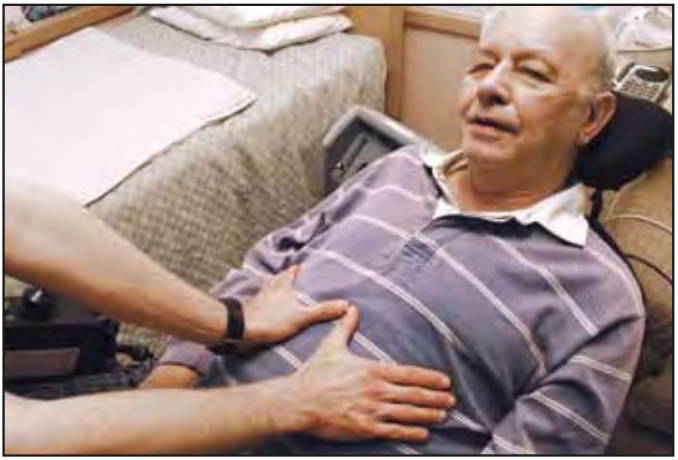
Some of the muscles you use in breathing are needed for coughing. Your doctor can tell you if your ability to cough isn't sufficient to clear secretions from your lungs and trachea.

The cough reflex is an explosive expulsion of a volume of air from the lungs. The high pressure and speed of this air propel irritants, such as mucus, up and out of the lungs. Neuromuscular disorders can weaken the ability to cough, but coughing is needed to remove mucus, especially during an acute respiratory infection.

The CoughAssist delivers air to the lungs and then quickly reverses air flow to pull out secretions.

A simple way to increase the effectiveness of your cough is a maneuver known as a *manual cough assist*. This maneuver involves a caregiver's pressing on the upper abdomen as you cough. A respiratory therapist or other health care professional can show you how to do this.

A cough assistance method that's effective even when cough muscles are severely weakened is a small electrical



A caregiver can deliver manual cough assistance by pressing on the upper abdomen, just beneath the ribs, as the patient coughs.

machine that first delivers a large volume of air and then quickly reverses air flow to pull out secretions. A brand name for such a device is the CoughAssist.

TREAT INFECTIONS

Monitor and treat respiratory infections promptly.

Once an infection starts, poor air exchange and weak coughing muscles make it difficult to clear mucus from your lungs. The mucus can become thick and can plug the airways, which can cause areas of the lung to collapse. (The medical term for lung collapse is *atelectasis*.)

Your doctor may suggest a chest X-ray for accurate determination of the presence of pneumonia or atelectasis.

Antibiotics will likely be prescribed if the infection is bacterial.

Cough assistance, either manual or via the CoughAssist, may be needed.

Some situations respond well to *high-frequency chest wall oscillation*, a form of vibration therapy delivered through a vest that shakes up mucus and moves it up where it can be coughed out.

WATCH FOR SCOLIOSIS

Scoliosis — a sideways curvature of the spine — is a common complication in neuromuscular disorders. Scoliosis prevents full expansion of the chest and can interfere with breathing.

Scoliosis in these diseases occurs because of weakening of the muscles that normally support the spine. At MDA clinic visits, especially in growing children, the spine will be physically examined for curvature, and the degree of the curve may be measured by X-ray. A curvature may progress surprisingly quickly in a child.

Keep a close eye on a progressing spinal curvature, which may crowd the lungs, and consider spine-straightening surgery on your doctor's recommendation.

IDENTIFY UNDERVENTILATION

The first consequence of weakened respiratory muscles usually is inadequate breathing during sleep. This underventilation occurs at night because the natural urge to breathe is lower during sleep, and because the abdomen pushes up against the diaphragm when you lie down.

The most common symptoms indicating insufficient breathing (*underventilation* or *hypoventilation*) are fatigue, poor sleep, nightmares or night terrors, and headaches, especially right after waking.

For many people with neuromuscular diseases, the beginnings of ventilatory failure come on slowly and may be mistaken for other problems. Shortness of breath — the best-known symptom of too little oxygen — may not occur if skeletal muscle weakness prevents you from exercising.

It's important to watch for the other signs of possible respiratory failure. These may include anxiety, confusion, loss of appetite and weight loss, weakening of the voice and weak coughing that doesn't move mucus up toward the mouth.

SYMPTOMS OF CHRONIC UNDERVENTILATION

- fatigue
- sleep disturbances
- nightmares, night terrors
- morning headaches
- confusion, disorientation, anxiety
- poor appetite, weight loss
- weakened or softened voice
- weak cough

PROPERLY TREAT SLEEP APNEA

Some studies have shown a high incidence of sleep disorders in people with neuromuscular diseases. But the cause of these problems may be different than for those without muscle diseases, and the treatment is different as well.

Among the general population, common causes of respiratory problems during sleep include pauses in breathing (*apnea*) either caused by brain abnormalities (called *central apnea*), or by collapse of the upper airway, blocking breathing (called *obstructive apnea*).

But for those with neuromuscular diseases, breathing problems during sleep may be caused or complicated by the fact that the muscles that aid respiration — the diaphragm and intercostals — have been weakened by muscle disease.

Weak respiratory muscles can lead to *nocturnal hypoventilation* (ineffective breathing during sleep) or *nocturnal apnea* (periodic cessation of breathing during sleep), when gravity, body position and neurological factors naturally make breathing efforts less effective.

A simple method to assess nighttime breathing difficulties is to measure exhaled carbon dioxide in combination with pulse oximetry, which painlessly measures blood oxygen levels through the placement of a small clip on a finger or toe. Typically these tests may be done at home while the patient sleeps.

A more sophisticated assessment tool is a sleep study or *polysomnogram (PSG)*, which pinpoints the causes of disrupted sleep through a combination of measurements, including encephalographic (brain) activity, eye movement, muscle activity, heart rhythm, respiratory effort and others. Polysomnograms are performed in a sleep laboratory and last between seven and 12 hours.

For the general population, continuous positive airway pressure, or CPAP, is the ventilation therapy commonly prescribed for obstructive apneas. CPAP blows in a continuous flow of air at a set pressure, keeping the airway from collapsing and obstructing breathing.

But CPAP often isn't appropriate for people whose problem is caused by weak respiratory muscles, because the muscles have to work harder to exhale against the constant inward flow of air.

In those cases, bilevel positive airway pressure ventilation, typically called BiPAP, is more commonly used. (BiPAP is a registered trademark of Respironics.) Air is administered at a higher pressure level on inhalation and a lower (or zero) pressure on exhalation.

In muscle diseases, the BiPAP "span," or difference between the inhalation and exhalation airflow pressures, is typically high to provide greater assistance to the inspiratory muscles and little or no resistance during exhalation.

Because sleep-breathing problems can be an effect of neuromuscular diseases, it's important to seek help at the first symptoms of underventilation. Be sure to consult an experienced, certified sleep specialist who knows which therapeutic solutions are appropriate for people with neuromuscular diseases.

RESPIRATORY TREATMENTS

There may be times when it's beneficial to deliver medication directly into the lungs.

Aerosol therapy is sometimes prescribed, especially to treat an infection or asthma that may be complicating your neuromuscular disease.

Medications can be delivered directly to the lungs with a nebulizer. This one is attached to a mouthpiece.

Nebulized medications, as well as air under pressure (see page 25), also can be delivered via a variety of masks, including some that appeal to children.

Some of the medications your doctor may prescribe include:

- mucolytics, which break down thick mucus
- decongestants, which decrease swollen tissues
- antibiotics, which combat infections
- bronchodilators, which open the airways and may assist with airway clearance

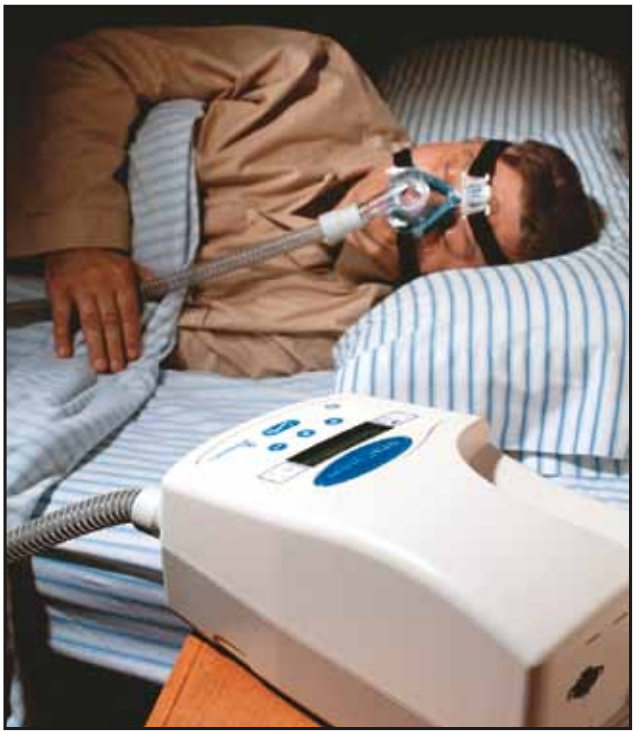
A nebulizer turns a liquid medication into a fine mist that can be inhaled. A small air compressor is attached to the nebulizer to generate a flow of air. The user breathes slowly and deeply through the nebulizer for 15 to 20 minutes three to four times a day or as prescribed.

When respiratory muscles have clearly been weakened, it's time to discuss *assisted ventilation* with your doctor and a respiratory therapist.

ASSISTED VENTILATION

Your doctor may have recommended that you consider assisted ventilation for yourself or your child when a neuromuscular disorder, such as muscular dystrophy or spinal muscular atrophy, has weakened respiratory muscles to the point that your own efforts can't provide adequate air exchange.

Assisting these muscles with mechanical support is in some ways similar to assisting skeletal muscles with a brace or heart muscle with a pacemaker.



Positive pressure devices pump air into the lungs under pressure. They're commonly used in neuromuscular diseases.

A belt that rhythmically inflates and deflates can help push air in and out of the lungs.

WHAT KINDS OF VENTILATORY ASSISTANCE ARE THERE?

If you're old enough to remember the polio epidemics of the 1940s and 1950s, you probably remember the term *iron lung*. These were in fact the first effective form of long-term ventilatory assistance, and they used a type of ventilation known as *negative pressure*.

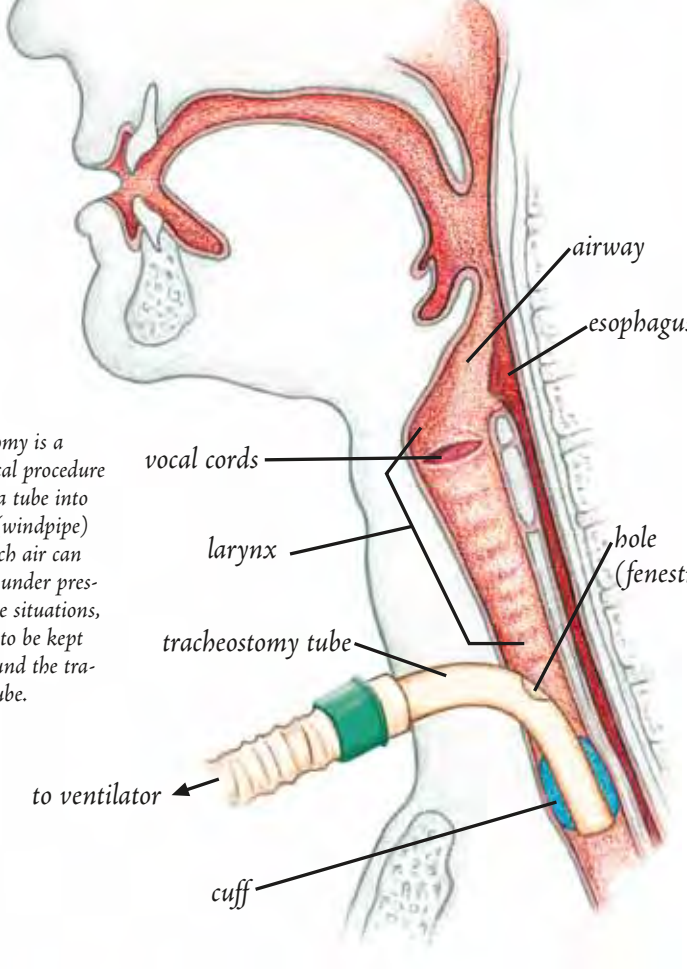
For the inspiratory part of the breathing cycle, the machine created a vacuum around the patient, who was encased in a metal tube (iron lung), pulling air into the lungs. The vacuum was released to allow the patient to exhale.

This type of device still exists, in an updated and much more portable form. A plastic shell that goes around the body, and can be worn while working, studying or sleeping, is a negative pressure type of assisted ventilation.

Another variation on this theme is a belt that rhythmically inflates — pushing against the diaphragm and aiding exhalation — and deflates, allowing chest expansion and aiding inhalation.

Today, though, devices that use *positive pressure* are much more common. With these devices, air from your surroundings is pumped into the lungs under pressure to simulate inhalation, with a lower pressure or no pressure during exhalation.

Iron lungs were used during the polio epidemics of the 1940s and 1950s. Photo courtesy of the March of Dimes.



A tracheostomy is a minor surgical procedure that inserts a tube into the trachea (windpipe) through which air can be delivered under pressure. In some situations, a cuff needs to be kept inflated around the tracheostomy tube.

Positive pressure devices can deliver either a set *volume*, or amount, of air, with variable pressures. Or they can deliver a set *pressure* of air, with variable volumes. These are generally called *volume-cycled* and *pressure-cycled* ventilatory assistance devices.

Volume ventilators typically (but not always) deliver air though a surgical opening in the *trachea* (windpipe), which provides more direct and controllable access to the lungs. This is called *invasive,* or *tracheostomy-delivered,* ventilation.

Volume-cycled ventilators are more powerful than pressure-cycled ventilators. They can be used in people whose respiratory muscles are extremely weak.

Pressure ventilators are smaller and less powerful. They typically deliver one pressure of air for inhalation and another for exhalation, so they're called *bilevel positive airway pressure*, or *BiPAP,* devices. (Some people don't call these ventilators at all, because they were originally used for other purposes.)

After a tracheostomy, a tube connects the windpipe to a mechanical ventilator.

BiPAPs usually are used without a tracheostomy, although doctors sometimes prescribe a small, pressure-cycled device with a tracheostomy for a young child with small lungs.

Adolescents and adults generally use a nasal mask, mouthpiece or other *noninvasive* (nontracheostomy) interface with these less powerful ventilators. People who use a mouthpiece requiring conscious effort will need to switch to a mask or other interface during sleep.

Today's ventilators are highly portable, battery-operated devices.

Ventilatory assistance need not be full time. In fact, many people use it only during sleep at first, progressing to daytime use gradually, if necessary.

Most people whose respiratory muscles are weak enough to require assisted ventilation also require assisted coughing. The CoughAssist (see page 16) or a similar device is often recommended, not only

COMPARISON: NONINVASIVE & INVASIVE ASSISTED VENTILATION

Factor	NoninvasiveSystem	Invasive System
Mechanism	air delivered through mouth or nose or by device around body	air delivered to lungs below mouth and nose through opening in windpipe (trachea)
Air exchange	effective for most until respiratory muscles are extremely weak	effective even when respiratory muscles are nonfunctional
Protection of airway	effective unless swallowing muscles are too weak to prevent inhalation of liquids or solids or when upper airway is obstructed	effective for all if tracheostomy tube has a cuff that protects airway from liquids or solids
Cost	generally covered by Medicare Part B or comparable insurance; some disposable supplies may require out-of-pocket payment	generally covered by Medicare Part B or comparable insurance; out-of-pocket costs for disposable supplies about $300 to $500 a month; additional long-term costs for professional caregivers
Surgery	no	yes (minor)
Control by user	adult user controls when to use system, switch to invasive system or use no system	user may lose ability to switch to noninvasive system
Infection risk	no increased risk of infection if device cleaned and maintained	increased risk
Inconvenience	minimal; can easily be used part time	need for suctioning of mucus, professional assistance, equipment sterility
Coughing	user can cough, with or without mechanical or manual assistance	will require coughing assistance or suction device to remove secretions from respiratory tract
Speaking	minimal interference, depending on interface	may interfere, especially when speaking valve can't be used
Swallowing	doesn't interfere	may interfere
Appearance	masks and other interfaces highly visible	face isn't affected; trach tube can be camouflaged

during colds but on a regular basis, to prevent mucus buildup in the airways.

WHY DO THESE DEVICES USE AIR INSTEAD OF OXYGEN?

There are some diseases — mainly diseases of the lungs themselves, such as cystic fibrosis and chronic obstructive pulmonary disease — in which a low and continuous flow of oxygen is beneficial. In these diseases, the air sacs of the lungs themselves are damaged, so that oxygen has a hard time flowing across them into the bloodstream, even though the person is moving air in and out of the lungs in a normal breathing cycle. Giving more oxygen with each inhalation can help in this situation.

But in neuromuscular disease, the primary problem (unless you have a pulmonary condition in addition to a neuromuscular one) is mechanical, making breathing too shallow. What's needed is help moving air in and out.

So, except during pneumonia or other acute illnesses, doctors usually prescribe nothing but room air — which is about 21 percent oxygen — for people with neuromuscular diseases who need ventilation.

INVASIVE OR NONINVASIVE VENTILATION?

Compared to invasive ventilation, noninvasive systems preserve natural swallowing, clearance of mucus and speaking. They don't have any surgical openings, so they don't lead to infections.

With a tracheostomy tube (trach) in place, mucus usually has to be suctioned out of the trach tube several times a day. The patient is vulnerable to infection because of the surgically created opening into the respiratory tract. Speaking can usually be maintained (if the person was able to speak before the trach), but it may not be completely normal. Swallowing also can be affected.

Some people with mild respiratory impairment find singing or playing a wind instrument helps their air exchange.

One way in which invasive systems may be superior to noninvasive is that they leave the face free of a visible device. Noninvasive systems require a visible mask, mouthpiece or other interface that some people find unacceptable.

HOW DO YOU SPEAK WITH A TRACHEOSTOMY TUBE IN PLACE?

During natural speech or singing, it's impossible to inhale. To produce sound, air has to move from the

lungs up the trachea and over the vocal cords in the larynx (see illustration, page 25).

For a trach user to talk, he or she has to be able to tolerate this temporary diversion of air away from the lungs. If you can tolerate this, a speaking valve (sometimes called a Passy-Muir valve, which is a brand name) can be attached to the trach tube. When you speak, this valve diverts air up over the vocal cords instead of in and out through the trach tube.

For air to move up to the vocal cords, there also has to be an opening in or around the trach tube. A hole in the upper surface of the tube or a deflatable cuff around the tube are two ways to allow this air movement.

CAN YOU SWITCH BETWEEN NONINVASIVE AND INVASIVE SYSTEMS?

It's a simple matter to switch from noninvasive to invasive ventilation. Switching from an invasive system to a noninvasive one is much more difficult, but it too can sometimes be accomplished. See the "Dear Friends" message on page 6.

RESPIRATORY CARE IN DUCHENNE MUSCULAR DYSTROPHY

By the time a boy with Duchenne muscular dystrophy (DMD) is in his teens — and sometimes earlier — he'll likely experience some respiratory distress, although this may not be recognized for what it is. Interrupted nighttime sleep, excessive daytime fatigue, headaches, and trouble concentrating and thinking may be due to poor air exchange. (See "Symptoms of Chronic Underventilation," page 19.)

In Duchenne MD and some other muscular dystrophies, it's important to realize that cardiac function is closely tied to respiratory function. A deteriorating heart muscle, also common in DMD and other MDs, can have a negative effect on breathing.

The relationship between heart function and breathing also works the other way: Respiratory abnormalities can contribute to cardiovascular problems. Doctors who study cardiomyopathy in muscular dystrophy say that using noninvasive assisted ventilation, particularly at night, helps heart function in boys with DMD.

Needless to say, poorly functioning cardiac and respiratory systems can be life-threatening. Early diagnosis, regular follow-up and early therapy for the heart or breathing problems can extend a young man's quality of life and length of life.

In 2004, the American Thoracic Society published a set of guidelines for respiratory care in DMD. The development of these guidelines was supported in part by MDA, through a grant to Jonathan Finder in the Department of Pulmonology at Children's Hospital of Pittsburgh.

The guidelines were written for physicians, but it's wise for parents and young men with DMD to be aware of them, and be able to discuss them with their physicians. One of the most important recommendations is to learn about assisted ventilation options before an emergency occurs.

The guidelines also recommend that a person with DMD:

- Undergo an evaluation of respiratory status early (between ages 4 and 6) and tests of respiratory function at every clinic visit thereafter.

- Undergo regular cardiac evaluations starting at school age.
- See a pulmonologist twice a year starting about age 12 or when FVC deteriorates to 80 percent of normal or less.
- See a pulmonologist every three to six months after starting to use assisted ventilation or an airway clearance device.

A child with Duchenne muscular dystrophy should begin having respiratory evaluations between ages 4 and 6.

- Have cough effectiveness evaluated regularly, and learn how to use a manual cough assist maneuver or an assisted coughing device.
- Learn how to use an oximeter at home to measure blood oxygen levels.
- Maintain good nutrition, with a feeding (gastrostomy) tube if necessary.

The experts strongly suggest that boys with DMD begin using noninvasive ventilatory support at night when breathing becomes insufficient during sleep or overnight blood oxygen measurements are below normal. When daytime breathing becomes inadequate, they should add noninvasive ventilation during the day.

When these methods begin to lose effectiveness, the young man and the family should consider the option of a tracheostomy.

The guidelines warn that supplemental oxygen shouldn't be used to treat inadequate breathing during sleep unless ventilatory assistance is also being used. And they caution against starting mechanical ventilation before it's required. Doing so can interfere with adequate respiratory monitoring and lead to a false sense of respiratory security.

RESPIRATORY CARE IN ALS

In most people with amyotrophic lateral sclerosis (ALS), respiratory muscles deteriorate as the disease progresses, because the nerve cells that control these muscles are lost. This deterioration leads to decreased air exchange. The results usually include shortness of breath when the person is lying down or with exertion, and interference with sleep — all signs of respiratory distress.

Another effect of weak intercostal muscles in ALS is a decrease in the person's ability to cough; infections can result. Manual or mechanical assisted coughing is usually recommended.

Regular measurements of respiratory function are crucial for people with ALS.

In addition, the nerves and muscles that control swallowing, as well as routing of food and liquids into the stomach and air into the lungs, are often affected. This can lead to inhaling of food or liquids, with resulting airway obstruction or infection. This type of problem may be solved by inserting a feeding (gastrostomy) tube so that the person isn't eating and drinking by mouth.

In 1999, the American Academy of Neurology published guidelines for physicians providing respiratory care in ALS. People with ALS can follow these recommendations and discuss them with their doctors.

- Watch for and report signs of respiratory muscle loss and decreased air exchange (see "Symptoms of Chronic Underventilation," page 19).
- Undergo frequent measurements of respiratory function.
- Begin noninvasive ventilation to relieve symptoms and to some extent prolong survival.
- Begin invasive (tracheostomy-delivered) ventilation if long-term survival is the goal, taking into account the burdens and benefits of this decision (see page 28).

The guidelines also advise doctors that the ALS patient maintains the right to refuse or withdraw treatment, including mechanical ventilation. While ventilation is being withdrawn, doctors should provide access to adequate pain medications (including morphine) and anti-anxiety drugs.

QUALITY OF LIFE

How will respiratory problems affect my quality of life? People with neuromuscular diseases who are using either invasive or noninvasive ventilation go to school and work; they go out with friends and travel. In almost all cases, speaking valves (Passy-Muir valves are one brand) now in use with trachs can redirect air flow while you're talking and allow your voice to be heard.

Trachs can be covered with stylish scarves.

Like most electronic equipment, vents have become smaller with time, and they can easily attach to a power wheelchair and accompany you almost anywhere. While you need to have access to care if your ventilator malfunctions, you're by no means confined to home.

The relief that vent users experience when they're no longer fighting for breath generally seems a fair trade for any self-consciousness about their appearance.

Many women with trachs have incorporated stylish scarves into their fashion decisions.

Early in the course of a neuromuscular disease, some people feel sure they'd never want to be on a breathing machine. But by the time you or your child needs ventilatory assistance, your feelings about what's necessary for a worthwhile life may have changed. You may find that being able to write poetry, paint pictures, manage your business or watch your children grow are well worth putting up with the inconvenience of a ventilator.

Assisted ventilation can help people forget the effort to breathe and concentrate on enjoying their families and activities.

In addition, future changes in technology are likely to produce ventilation equipment that's even smaller, lighter and more effective than today's devices.

In 2002, a study of 26 ventilator users in Canada found that participants reported they had a good quality of life. Health professionals and the general public generally saw ventilation as an intrusive burden, while ventilator users saw it as assistive technology.

The decision to use ventilation and what kind to use depends on many factors, and medical issues aren't the only ones to be considered. When making the choice, it's worth keeping in mind that people (even professionals) who haven't themselves used ventilatory assistance probably don't know how you or your child will experience it.

ADVICE FROM A VETERAN VENT USER

Tedde Scharf, who has muscular dystrophy and has used assisted ventilation since 1988, offers this advice.

Emergency squeeze bags

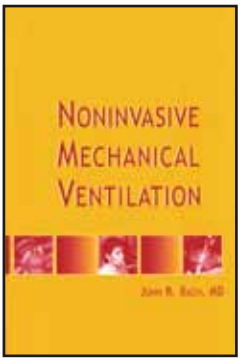
- Know and understand the signs of respiratory insufficiency and seek medical help early.
- If you need assisted ventilation, learn about all the options, their benefits and drawbacks.
- Learn everything about your medical needs and the equipment you use, and plan an organized routine for your caregivers.
- Select a clean, dedicated location for all equipment and supplies. Keep it well organized so that caregivers know exactly where to find everything.
- Keep an emergency squeeze bag attached to a face mask (resuscitator) nearby in case of equipment failure.

Tedde Scharf with her sister and business partner, in their new bookstore in Tubac, Ariz.

RESOURCES

BOOKS

Long-Term Mechanical Ventilation, ed. by Nicholas Hill, M.D., 2001, Marcel Dekker



Management of Patients With Neuromuscular Disease, by John Bach, M.D., 2004, Elsevier Health Sciences Division, www.us.elsevierhealth.com

Noninvasive Mechanical Ventilation, by John Bach, M.D., 2002, Elsevier Health Sciences Division, www.us.elsevierhealth.com

2006 Resource Directory for Ventilator-Assisted Living, in print and online, International Ventilator Users Network (IVUN) Division, www.post-polio.org/ivun. Also publishes "Ventilator-Assisted Living," quarterly newsletter

MDA

The following MDA publications can be found on the MDA website, www.mda.org, or through your local MDA office. Quest is MDA's quarterly national magazine. MDA publishes the MDA/ALS Newsmagazine for those with ALS (amyotrophic lateral sclerosis, or Lou Gehrig's disease).

QUEST

"Getting a Tracheostomy: My Story," April-June 2011

"NIV Masks — Finding the One That's Just Right," March-April 2008

"Not Enough ZZZzzzs," sleep-breathing problems, March-April 2008

"Truth, Lies and Tracheostomies," July-August 2007

"Hip to be Trach Chic," November-December 2005

"The Great Trach Escape," noninvasive ventilation, September-October 2003


The Great
Trach Escape —
IS IT FOR YOU?
Portable but Powerful Equipment, Skilled Guidance Make Noninvasive Ventilation an Option for Many
by Margaret Wahl
Adamant About Another Way
Life With a BiPAP

"Staying Ahead of the Curve," scoliosis surgery, August 2002

"Better Nights for Better Days," sleep problems, October 2000

"A Breath of Fresh Air: Respiratory Care Can Improve Quality of Life," December 1998

"Breathe Easy: Options Offered for Respiratory Care," October 1998


Chapter 5
Respiratory Issues

MDA ALS DIVISION

Everyday Life With ALS, 2005, Chapter 5, "Respiratory Issues"

MDA ALS Caregiver's Guide, 2008, Chapter 3, "Respiratory Issues"

"Managing Mucus Plugs," June 2009

"A Tale of Two Vent Choices," MDA/ALS Newsmagazine, August 2003

"Noninvasive Ventilation Prolongs Life if Used Right," MDA/ALS Newsmagazine, September 2002

"Noninvasive Ventilation Can't Sustain Life Indefinitely in ALS," MDA/ALS Newsmagazine, April 2001

"What Everyone with ALS Should Know About Breathing," MDA/ALS Newsmagazine, August 2000

MDA VIDEOS

"Breathe Easy: A Respiratory Guide for People Living With Neuromuscular Diseases"

"Breath of Life," for medical professionals

MDA

The Muscular Dystrophy Association fights neuromuscular diseases through an unparalleled worldwide research effort. The following diseases are included in MDA's program:

Muscular Dystrophies

Myotonic dystrophy *(Steinert disease)*
Duchenne muscular dystrophy
Becker muscular dystrophy
Limb-girdle muscular dystrophy
Facioscapulohumeral muscular dystrophy
Congenital muscular dystrophy
Oculopharyngeal muscular dystrophy
Distal muscular dystrophy
Emery-Dreifuss muscular dystrophy

Motor Neuron Diseases

Amyotrophic lateral sclerosis *(ALS)*
Infantile progressive spinal muscular atrophy *(Type 1, Werdnig-Hoffmann disease)*
Intermediate spinal muscular atrophy *(Type 2)*
Juvenile spinal muscular atrophy *(Type 3, Kugelberg-Welander disease)*
Adult spinal muscular atrophy *(Type 4)*
Spinal-bulbar muscular atrophy *(Kennedy disease)*

Inflammatory Myopathies

Polymyositis
Dermatomyositis
Inclusion-body myositis

Diseases of Neuromuscular Junction

Myasthenia gravis
Lambert-Eaton (myasthenic) syndrome
Congenital myasthenic syndromes

Diseases of Peripheral Nerve

Charcot-Marie-Tooth disease
Friedreich's ataxia
Dejerine-Sottas disease

Metabolic Diseases of Muscle

Phosphorylase deficiency *(McArdle disease)*
Acid maltase deficiency *(Pompe disease)*
Phosphofructokinase deficiency *(Tarui disease)*
Debrancher enzyme deficiency *(Cori* or *Forbes disease)*
Mitochondrial myopathy
Carnitine deficiency
Carnitine palmityl transferase deficiency
Phosphoglycerate kinase deficiency
Phosphoglycerate mutase deficiency
Lactate dehydrogenase deficiency
Myoadenylate deaminase deficiency

Myopathies Due to Endocrine Abnormalities

Hyperthyroid myopathy
Hypothyroid myopathy

Other Myopathies

Myotonia congenita
Paramyotonia congenita
Central core disease
Nemaline myopathy
Myotubular myopathy
Periodic paralysis

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