

BREATHING BASICS

Respiratory Care for Children with Spinal Muscular Atrophy



Contents

Introduction to Respiratory Care	4
• What is respiratory care?	4
• Why is respiratory care so important in SMA?	4
• How do we breathe normally?	4
• How do individuals with SMA breathe?	5
➢ Stomach breathing	
➢ Bell-shaped chest	
➢ Sunken chest	
• What are common breathing problems for children with SMA?	6
➢ Lung underdevelopment	
➢ Weak cough	
➢ Risk of infection and pneumonia	
➢ Viral infections, RSV	
➢ Swallowing problems, aspiration	7
➢ Sleep problems, hypoventilation	
Elements of Respiratory Care Management in SMA	8
#1: Assessment, Evaluation and Monitoring	8
#2: Breathing Exercises for Lung Development	9
➢ Resuscitator bag and mask	
➢ Cough machine	
#3: Coughing Practice for Airway Clearance	10
➢ Cough machine	
➢ Manual cough assist	
#4: Removal of Mucus from Airways and Lungs (Secretion Mobilization)	11
➢ Chest physiotherapy (manual/mechanical percussion)	
➢ Postural drainage	12
➢ Intrapulmonary percussive ventilation (IPV)	
➢ High frequency chest-wall oscillation or vest therapy	
#5: Measuring Oxygen Levels	12
➢ Pulse oximetry	
#6: Breathing Support Options	13
➢ Non-invasive respiratory management	
➢ Invasive respiratory management	



Who Should Read this Booklet?

This booklet is written for parents and families of children with Spinal Muscular Atrophy (SMA). Health care professionals involved in SMA care, and the general public, may also find this information very helpful.

➤ Night-time breathing support options	14
➤ Bi-level positive airway pressure (BiPAP)	14
➤ Mechanical ventilator	15
➤ Negative pressure ventilation (NPV)	15
➤ Sipper Vent	16
#7: Care During a Cold	16
➤ Evaluation of mucus build-up and secretion mobilization	16
➤ Cough machine	17
➤ Breathing support	17
➤ Pulse oximeter and oxygen saturation	17
➤ Dehydration	17
➤ Cold prevention, RSV	18
#8: Perioperative Evaluation (Before and After Surgery)	18
Special Needs of Children with SMA	19
• SMA Type I	19
➤ Breathing support	19
➤ Respiratory care options and difficult decisions	20
➤ Non-invasive respiratory care	20
➤ Invasive respiratory care with intubation	21
➤ Palliative care	22
• SMA Type II	23
➤ Care during a cold	23
• SMA Type III	24
Conclusion	25
• Breathing Basics Summary	25
• About Families of SMA	28
• For more information	29
• Respiratory equipment at home	29
• For copies of this booklet	30
• Disclaimer	30



Author:

Mary K. Schroth, M.D.
Associate Professor of Pediatric Pulmonology
Director, Pediatric Pulmonology Center Grant
American Family Children's Hospital
University of Wisconsin School of Medicine and Public Health

Expert Review Panel:

- Richard S. Finkel, M.D.
- Albert M. Freedman, Ph.D.
- John T. Kissel, M.D.
- Richard M. Kravitz, M.D.
- Kristin J. Krosschell, M.A., P.T.
- Kathryn J. Swoboda, M.D.

Editors:

- Marilyn Weisberg, M.P.H.
- Lindsey Harle, M.D.

Funding:

- The Angel Baby Foundation and Families of SMA.

What is Respiratory Care?

Respiratory care deals with lung function and health, including breathing.

Pulmonary means *relating to the lungs*. *Pulmonary medicine* and *pulmonology* are also fields that deal with diseases of the lungs and respiratory tract. In some countries, these disciplines are called *chest medicine* and *respiratory medicine*.

Pulmonologists (lung specialists) and respiratory care therapists help people with breathing problems. A *pediatric pulmonologist* is a lung specialist who works closely with children.

Why is Respiratory Care So Important in SMA?

Respiratory (breathing) problems are the leading cause of illness for children with SMA. They are the most common cause of death for children with SMA Type I and SMA Type II. Respiratory care interventions are essential to the survival and comfort of children with SMA.

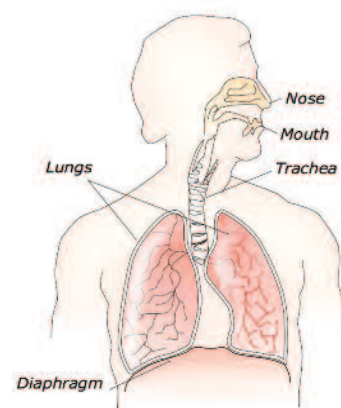
As a parent, it is very important that you watch for breathing problems in your child. You will want to talk with the doctors, including a pediatric pulmonary specialist, to develop respiratory care goals that are personalized for your child. *(from the Consensus Statement for the Standard of Care in SMA)*

How Do We Breathe Normally?

We breathe using two main muscle groups: the intercostal muscles and the diaphragm. The intercostal muscles are the muscles between the ribs. These muscles help the rib cage expand as the lungs fill (inflate) with air.

The diaphragm is a circular muscle located at the bottom of the rib cage. The diaphragm helps the rib cage to move down, allowing the lungs to fill (inflate) with air.

The intercostal muscles and the diaphragm work together in normal breathing. By expanding the chest, these muscles allow the lungs to fill with air during inspiration (when we breathe in). The oxygen in this air is then delivered from the lungs to the rest of the body through the bloodstream. During expiration (when we breathe out), carbon dioxide is released from the lungs into the air.



Location of diaphragm muscle in relation to the lungs.

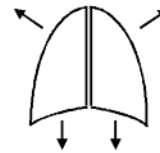
How Do Individuals with SMA Breathe?

Stomach Breathing

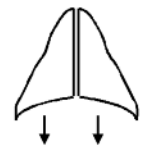
Children with SMA breathe differently. Their intercostal muscles between the ribs are weak. Their diaphragms are stronger and become the major muscle used for breathing. With weak intercostal muscles, the rib cage does not expand outward during breathing. The diaphragm, however, stays strong and pulls the rib cage downward. This causes what appears to be “stomach breathing”—when the child expands the stomach while breathing, instead of the chest.

Chest Wall Changes

Normal



SMA



(Courtesy of M. Schroth, M.D.)

Bell-Shaped Chest



Bell-shaped chest

Children with SMA may appear to have “bell-shaped” chests, which look wider at the bottom than at the top. This happens because the weak intercostal muscles do not help the upper chest to expand (or go out) normally during breathing, and the diaphragm pulls the chest and rib cage down.



Stomach Breathing



Sunken Chest or Pectus Excavatum

Sunken Chest or Pectus Excavatum

Children with SMA also develop “pectus excavatum.” This is when the sternum, the bone in the middle of the chest, appears to be sunken. When the diaphragm pulls the rib cage down, the sternum is also pulled in.

What are Common Respiratory Problems in Children with SMA?

Normally, the muscles in the rib cage are strong. They allow the lungs to take in oxygen and expel carbon dioxide efficiently. For children with SMA, the weak muscles in the upper chest make breathing much more difficult. As a result, they can have a number of respiratory problems including lung underdevelopment and weak cough; increased difficulty with infection, including viral infections and pneumonia; swallowing problems and aspiration; and sleep problems with hypoventilation.

Lung Underdevelopment

As children grow, their lungs should continue to grow and develop. For children with SMA, their weak intercostal muscles do not help the lungs to develop properly. Over time, their lungs do not grow to normal size and their chest muscles do not become as strong as we would like them to be.

Weak Cough

When a child with SMA develops a cold, flu, or viral respiratory infection, there is often an increase in secretions (mucus) from the nose and lungs. A strong cough helps to clear these secretions and prevent blockage of the airways.

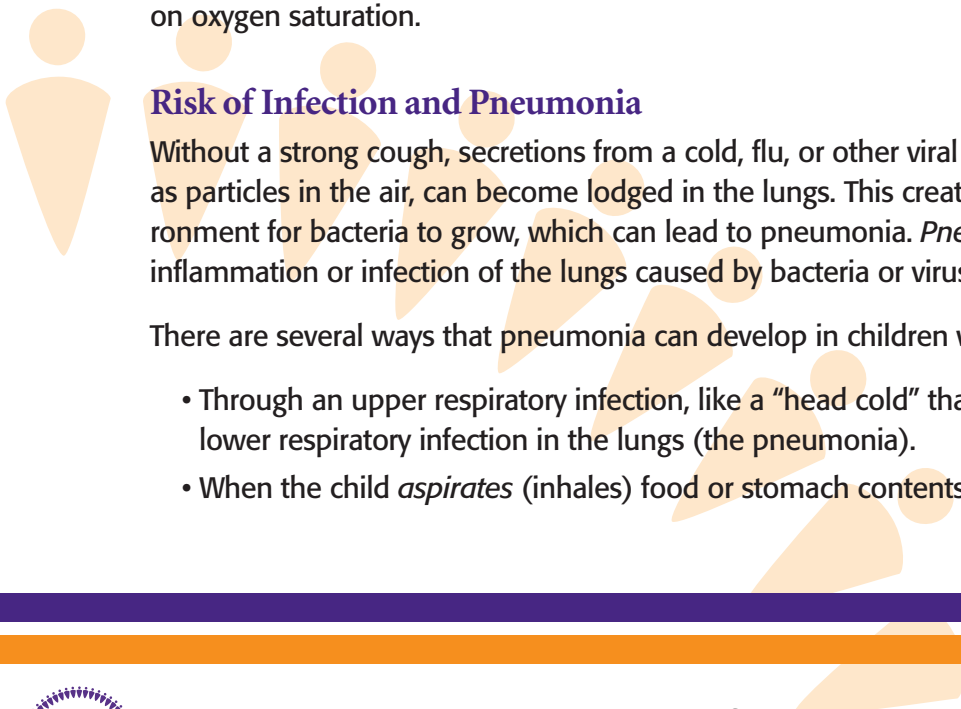
Children with SMA do not have enough muscle strength for a strong cough. With poor coughing, secretions stay in the lungs and compromise (decrease) breathing. When breathing tubes and airways become plugged, parts of the lungs can collapse resulting in a condition called *atelectasis*. Oxygen saturation levels (a measure of how much oxygen is in the blood) can also drop. See Section #7 for more on oxygen saturation.

Risk of Infection and Pneumonia

Without a strong cough, secretions from a cold, flu, or other viral infections, as well as particles in the air, can become lodged in the lungs. This creates an ideal environment for bacteria to grow, which can lead to pneumonia. *Pneumonia* means inflammation or infection of the lungs caused by bacteria or virus.

There are several ways that pneumonia can develop in children with SMA:

- Through an upper respiratory infection, like a “head cold” that turns into a lower respiratory infection in the lungs (the pneumonia).
- When the child *aspirates* (inhales) food or stomach contents into the lungs.



Viral Respiratory Infections, RSV

Viral respiratory infections can be life threatening. One example is RSV which stands for *respiratory syncytial virus*. This virus can look and sound like the common cold. Anyone can catch it and for most children RSV is no more than a bad cold. For children with SMA, however, RSV can be very serious and lead to severe breathing problems, even hospitalization.

Like the common cold, RSV is highly contagious and most often occurs in the late fall and winter months. It spreads very easily from person to person and by touching any objects, like toys, infected with the virus. There is a medication in injection form that your doctor may prescribe to try to prevent RSV. This is used only for a small number of babies at very high risk for hospitalization.

As a parent, you can help to prevent colds, flu, and RSV through some common-sense habits. These include good hand washing for everyone in the house; keeping infected siblings away from the child with SMA; and keeping your child away from busy, public places like malls and day care centers, during the cold season.

See Section #1 on Assessment, Evaluation, and Monitoring for more information on immunizations and vaccines; and Section #7: Care During A Cold. Be sure to talk with your doctors about prevention and treatment measures.

Swallowing Problems, Aspiration

Children with SMA often have problems with swallowing, which can cause them to *aspirate* (inhale) food into their lungs while eating. In addition, children with SMA also frequently have acid reflux or gastroesophageal reflux, and heartburn. Acid reflux happens when liquid or food in the stomach containing acid travels back from the stomach into the *esophagus* (swallowing tube), and up into the mouth. Heartburn is a burning feeling rising from the stomach or lower chest up to the neck. Poor swallowing due to muscle weakness can lead to inhaling the liquid or food that comes up from the stomach into the lungs while the child is breathing.

Food and other foreign particles in the lungs can lead to pneumonia. When pneumonia is due to breathing in food or liquids, this is called *aspiration pneumonia*.

Sleep Problems, Hypoventilation

Normally, the muscles of the body relax during sleep, including the muscles we use for breathing. For children with SMA, this relaxation of the breathing muscles can result in *hypoventilation*. Hypoventilation means that breathing is too shallow or too slow. The lungs are not taking in (inhaling) enough oxygen or giving off (exhaling) enough carbon dioxide to meet the needs of the body. Hypoventilation during sleep is often one of the earliest signs of breathing difficulty in SMA.



1 Assessment, Evaluation, and Monitoring

We recommend that all parents of children with SMA talk with their doctors about their child's respiratory care management. Each child is different and requires his or her own respiratory care plan with goals, including a plan for times of illness. These goals should reflect the needs of your child and what you think is best. The goals should also consider what is reasonable for you as a parent and caretaker, and for your family.

Children with SMA need regular evaluations from the doctor every 3 to 6 months, generally, to assess their breathing and look for any problems. Children with SMA Type III may need to be seen less often. The following recommendations are described for the 3 functional levels in SMA: Nonsitters, sitters, and walkers.

Nonsitters should be evaluated on a regular basis for:

- cough effectiveness
- gas exchange, including the level of oxygen in the blood
- chest wall deformities (changes)
- breathing during sleep, with polysomnography (sleep study)
- chest x-ray
- ability to swallow
- new or unexplained respiratory difficulty

Sitters should be monitored on a regular basis for:

- cough effectiveness
- chest wall deformities (changes)
- breathing during sleep, with polysomnography (sleep study)
- scoliosis

Walkers should be evaluated periodically for:

- cough effectiveness
- pulmonary function, with spirometry (test of breathing capacity).
- other tests, based on clinical symptoms and observations.

All children with SMA should receive regular vaccines, appropriate nutritional support, hydration, and monitoring for gastro-esophageal reflux (GERD).

For more information on immunizations and vaccines, see guidelines on the website of the American Academy of Pediatrics at <http://www.aap.org>. Be sure to talk with your doctors about medications and prevention techniques that may reduce the risk of your child developing colds, influenza (flu), and respiratory infections, including RSV. Also, see Section #7 on Care During A Cold.

Breathing Exercises for Lung Development

2

Breathing exercises, usually done at home, can help children with SMA develop their lungs. These interventions and exercises also help the children to improve their ability to more fully inflate their lungs.

Resuscitator Bag and Mask

The resuscitator bag and mask can help with breathing. If you place the mask over your child's nose and mouth, you can provide a breath with the bag as your child inhales. This can be repeated several times, for several minutes.



Resuscitator bag and mask



Cough assist machine, tubing, and face mask.

Cough Machine

A cough machine can be used to help your child inhale and exhale. The machine forces air into the lungs at a preset pressure and then sucks the air out of the lungs at a preset pressure.

For lung development, set the cough machine in the *inhalation only* mode by turning the exhale time to "0". Place the mask over the child's nose and mouth or use a mouthpiece. With a preset pressure, the machine will then help the child's lungs to inflate.



3 Coughing and Airway Clearance

Coughing (or airway clearance) is necessary to remove nasal secretions and inhaled particles. Helping children with SMA to cough more forcefully prevents lung infections and other respiratory complications associated with retained secretions. Coughing can be improved with a cough machine or with manual cough assistance.

Cough Machine

A cough machine can also be very helpful for coughing and airway clearance. As mentioned above, the machine pushes air into the lungs and then sucks the air out of the lungs at preset pressures. This action can help produce a more effective cough for your child, more like the cough of a healthy child. A typical pattern is to rest for 1 to 2 minutes between sets of 4 to 5 breaths. You can obtain a cough machine from a durable medical supply provider with a prescription from your doctor.



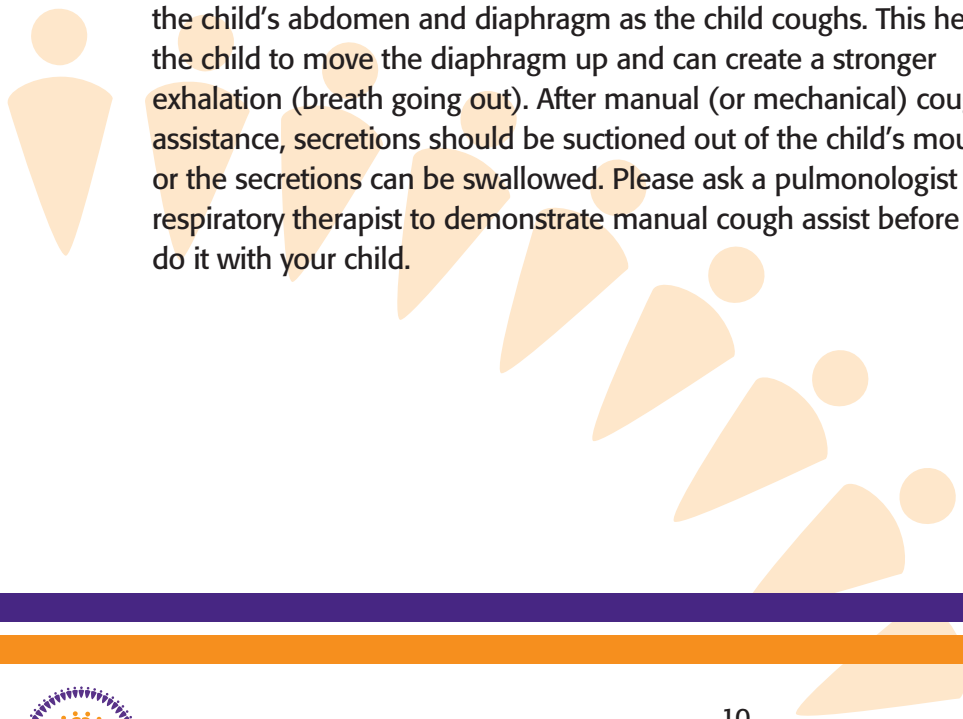
Cough machine

Manual Cough Assistance

Manual means *by hand*. In manual coughing assistance, a parent or other caretaker places gentle upward pressure with their hands on the child's abdomen and diaphragm as the child coughs. This helps the child to move the diaphragm up and can create a stronger exhalation (breath going out). After manual (or mechanical) cough assistance, secretions should be suctioned out of the child's mouth or the secretions can be swallowed. Please ask a pulmonologist or respiratory therapist to demonstrate manual cough assist before you do it with your child.



Cough assistance with intubation



Removal of Mucus from the Airways and Lungs (Secretion Mobilization)

4



Chest Physiotherapy with palm cups

During colds, most children with SMA have more trouble coughing. They are unable to effectively clear out the secretions that build up in their airways. There are several techniques available to help with secretion mobilization, or the removal of mucus from the airways and lungs. These techniques include chest physiotherapy (both manual and mechanical), postural drainage, intrapulmonary percussive ventilation (IPV), and high-frequency chest-wall oscillation (vest therapy).

Chest Physiotherapy (also known as Manual or Mechanical Percussion)

Chest physiotherapy is the gold standard for secretion mobilization. It is used to loosen secretions so they are easier to cough up. With the manual percussion (vibrating, clapping) technique, the caregiver uses his or her hands and gravity to help loosen secretions. You can use a face mask or palm cups to gently clap on the child's chest wall. Ask your healthcare provider if this is advisable for your child and to show you how to properly do chest physiotherapy.



Cupped hand for chest physiotherapy

In mechanical percussion, the caregiver places a vibrating device on the child's chest to loosen secretions.

Children who are severely affected by SMA may need cough assistance using chest physiotherapy on a daily basis. Others may only need assistance when they are sick.

Postural Drainage

In postural drainage, the child is positioned at an incline (pillows and wedges help) with his or her head and chest lower than the buttocks. Gravity then pulls secretions from the lower airways into the upper airways, where they can be removed by suctioning or coughing.



Postural Drainage



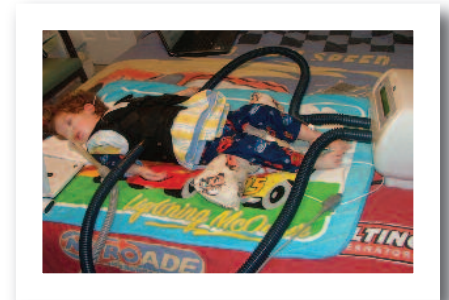
Intrapulmonary Percussive Ventilation (IPV)

An IPV is a machine that provides positive air pressure into the lungs at a set frequency and sounds like a choo-choo train. Medication or saline is nebulized (turned into a fine mist that travels with air) into the child's lungs. The pressure and frequency are preset to a comfortable level for the child. This shakes and loosens the secretions inside the lungs so that they are easier to cough out.

IPV is expensive and may be no more effective than other less expensive methods, like chest physiotherapy, for removing fluids from the lungs.

High Frequency Chest-Wall Oscillation or Vest Therapy

This device is worn like a vest. It gently shakes the chest and loosens the secretions inside the lungs. The vest is expensive and may be no more effective than the other less-expensive methods, like chest physiotherapy, for removing fluids from the lungs.



Vest Therapy

5 Measuring Oxygen Levels

Pulse Oximetry

Measurements of pulse oximetry (the amount of oxygen in the blood) can be performed with a simple, lightweight clamp or Band-Aid-like wrap on the child's finger or toe. This measurement will tell you if your child is clearing secretions adequately. If your child has very weak breathing in general, you may find it helpful to measure oxygen saturation (the oxygen level in the blood) using the pulse oximetry when he or she is well, so that you have a baseline measurement. You can then compare this measure from healthier times against the measure when ill. 95% or higher is considered normal oxygen saturation.

For more on pulse oximetry and oxygen saturation, see #7: Care During A Cold.



Breathing Support Options



Non-invasive breathing support with nose mask

Non-invasive Respiratory Care Management

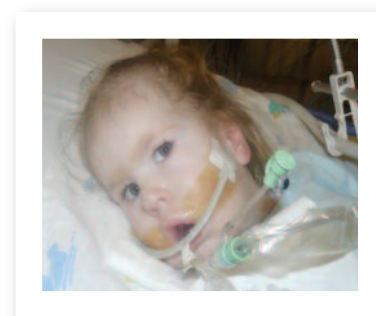
Non-invasive means that no devices are used that are inside or penetrate the body as part of the breathing procedure. Everything is on or outside the body.

Non-invasive respiratory care will help your child breathe using an air-tight mask over the nose, or nose and mouth. The mask is connected to a machine that helps with breathing.

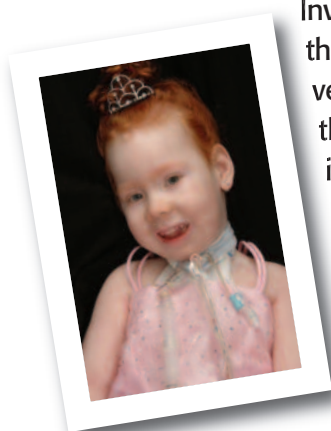
Invasive Respiratory Care Management

Invasive means that devices are used that are inside or penetrate the body to help with breathing. Needles, tubes, lines, and surgery may be used.

Invasive respiratory care will help your child breathe through a breathing tube attached to a mechanical ventilator (breathing machine). The tube goes into the body through the mouth or through a small hole in the neck, and into the lungs. At first, invasive respiratory care uses a breathing tube, called an *endotracheal tube*, through the mouth. The endotracheal tube, if left in the airway for more than a few days to weeks, can cause damage and irritate the mouth and throat. The placement of a tube into the body is called *intubation*.



Intubation with endotracheal tube



Invasive breathing support with a tracheostomy

When ventilator support is needed for an extended period of time, the doctor may need to place a tracheostomy tube. For the tracheostomy, the doctor makes a small, surgical hole in the child's neck. A breathing tube, called a "trach," is inserted through this hole, where it bypasses the mouth and vocal cords, and goes directly into the large airway, known as the trachea.

Night-time Breathing Support Options

Children with SMA Type I, and some children with Type II, have very weak breathing during sleep, with short and shallow breaths, and poor oxygen and carbon dioxide exchange (hypoventilation). When this happens, the child has too little oxygen and too much carbon dioxide in the body.

When oxygen levels are too low and carbon dioxide levels are too high, the body cannot function normally. To prevent hypoventilation, some children need mechanical breathing support while they are sleeping to help them rest their breathing muscles. If they become sick with a cold or flu, they may need this support even while awake.

With improvements in oxygen exchange and better sleeping, children with hypoventilation may have less nighttime sweating, fewer headaches, better appetites and weight gain, and better concentration.

There are 3 ways to help children with SMA breathe while sleeping. These include use of BiPAP, mechanical ventilation, and negative pressure ventilation (NPV).

Bi-level Positive Airway Pressure (BiPAP, pronounced “bye-pap”)

The BiPAP machine provides two levels of positive airway pressure delivered through a mask over the nose, or over the nose and mouth, or through a tube under the nose and into the nostrils. The machine provides higher pressure and an increased volume of air when the child inhales. When the child exhales, the machine lowers its pressure automatically to allow for a more normal breathing pattern.

A BiPAP machine can sense a child’s normal breathing cycle and synchronize (work at same time or rate) with this cycle. The machine will also give breaths when the child is in deep sleep and is not breathing adequately on his or her own.

These are the BiPAP settings commonly used in SMA care that help children to rest their breathing muscles:

- Inspiratory positive airway pressure (IPAP) of 14 to 20 cm H₂O.
- Expiratory positive airway pressure (EPAP) of 3 to 6 cm H₂O.
- Respiratory rate of 14 to 30 breaths per minute, based on age and resting respiratory rate.
- Inspiratory time of 0.5 to 1.5 seconds, based on age and respiratory rate.

Continuous positive airway pressure (CPAP) is a different type of setting that gives one continuous amount or level of pressure. This may not provide adequate rest for breathing muscles. It is not recommended for SMA patients.



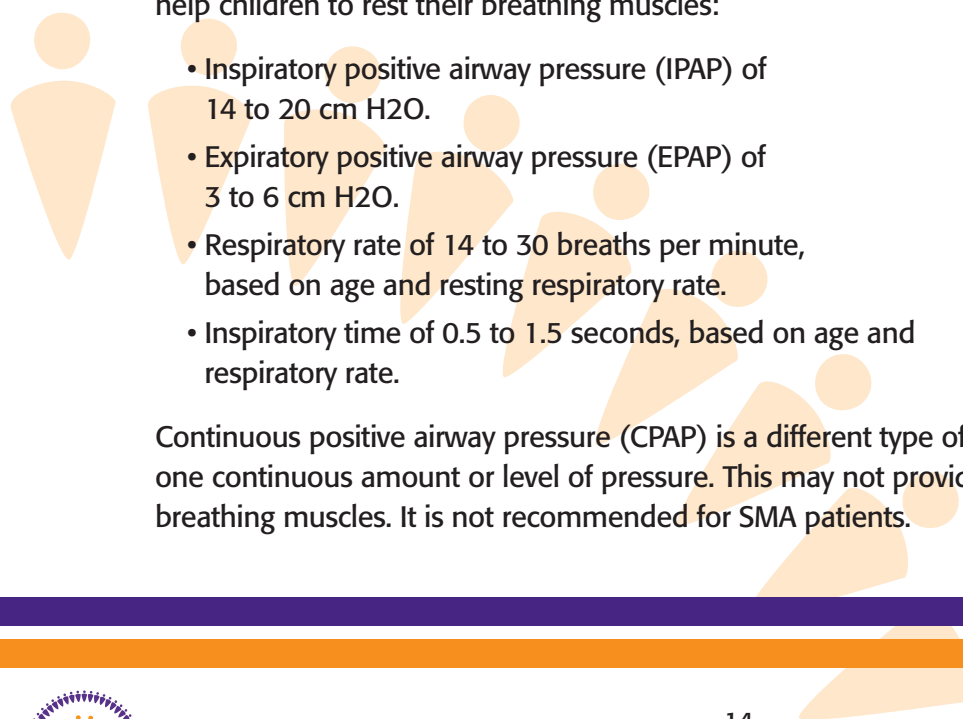
BiPAP with nasal prongs



Bipap with nose mask



Child on supplemental oxygen with NG (nasogastric) tube





Mechanical ventilator

Mechanical Ventilator

Mechanical ventilators have more options and settings than BiPAP and they allow for better control of breathing. Mechanical ventilation can be delivered using a nose mask, a nose/mouth mask, or through a tracheostomy tube. This type of breathing support can be used while the child is awake and/or asleep.

There are different types of mechanical ventilators, including some that can be carried over the shoulder or placed on a wheelchair. Portable external batteries are also available.

Negative Pressure Ventilator (NPV)

This type of ventilator encircles the chest in a large, external chamber, much like the “iron lung” used many years ago. The chamber is connected to a vacuum that draws air out of the chamber. As air goes out, negative pressure in the chamber builds up, helping to expand the chest and bring air into the lungs. The ventilator can be set to a specific vacuum pressure and respiratory rate. A Porta-Lung is an example of a negative pressure ventilator. Some children continue to use NPV, but this type of ventilation is rarely prescribed today due to technological improvements in BiPAP and mechanical ventilators.

Sipper Vent

This device allows a child or adult to take a breath while in the wheelchair through a small mouthpiece, positioned like a microphone in front of the face. The mouthpiece is connected to a ventilator which provides a boost of air for talking and breathing at the same time.



sipper vent

7 Care During a Cold (upper respiratory illness)

Evaluation of Mucus Build-up and Secretion Mobilization

During a cold, children with SMA may have difficulty clearing mucus from their lungs. To reduce this problem, take these steps in this order every 4 hours, and as needed, during a cold:

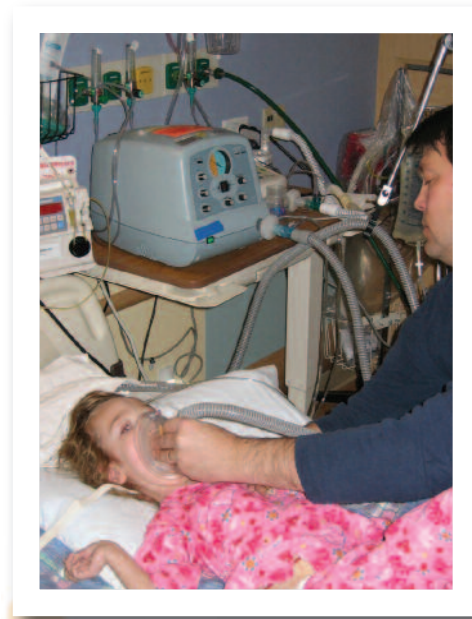
- *Airway secretion mobilization*: Do this for 10-20 minutes. Use a technique that loosens secretions, including chest physiotherapy with your hands or with palm cups; or electric percussor (mechanical percussion); or high frequency chest-wall oscillation (vest therapy); or intrapulmonary percussive ventilation (IPV).
- *Cough machine*: Do 4 sets of 5 breaths to remove the loosened secretions. Then, suction secretions from the child's mouth.
- *Postural drainage*: Do this for 15-30 minutes. Position the child at an incline (pillows and wedges help) with the child's head and chest lower than the buttocks.
- *Cough machine, again*: Do 4 sets of 5 breaths, again, to remove the loosened secretions. Then suction secretions from the mouth, again.

Cough Machine

The cough machine is an important tool and you can never use it too much! Use the cough machine every time your child's breathing sounds coarse or there is trouble coughing out secretions. If your child has difficulty coughing out secretions from the back of the throat, you may also suction the mouth.

Breathing Support

Colds make children with SMA weaker. During a cold, if your child is already on BiPAP or another breathing support, use the BiPAP during all sleep, including naps during the day. Breathing support may also be needed when your child is awake and sick with a cold. Use the pulse oximeter, as described in the next paragraph, to know when your child needs to be on BiPAP.



Child on cough machine



Pulse Oximeter and Oxygen Saturation

Use a pulse oximeter to check your child’s blood oxygen level when the child is well to establish a baseline. Then use it more frequently if your child appears to be having difficulty breathing. Use it often if your child is sick. It is not always easy to know if your child is having breathing difficulty just by looking. The pulse oximeter will tell you if there is a problem and if your child needs help with coughing.

Oxygen saturation is a measure of how much oxygen is in the blood. Children with SMA need extra breathing support when their oxygen saturation level drops too low.

General Guidelines for Oxygen Saturation

IF...	THEN...
If oxygen saturation is less than 94%...	Use the cough machine to clear secretions while monitoring the pulse oximeter.
If oxygen saturation is 92% or less and the cough machine is not increasing the oxygen saturation...	First, place your child on BiPAP, or another breathing support device, and give airway clearance treatment; then contact your physician.
If you cannot maintain oxygen saturation greater than 90% while on BiPAP and in-room air...	Call emergency services because your child may need to be in the hospital for more intense therapy.

Dehydration

If your child is vomiting, has diarrhea or a poor appetite and is not taking in fluids, he or she may become dehydrated. This is a serious complication in children with SMA. You should call your doctor if you see these symptoms and suspect any dehydration.

Cold Prevention, including RSV

There are important steps you can take to prevent exposure to colds, RSV, and other viruses, particularly in the first few months of your child’s life:

- Make sure everyone washes their hands before touching your child.
- Keep your child away from anyone who has a cold, fever, or runny nose.
- Keep your child away from crowded areas, like shopping malls.
- Keep your child away from tobacco smoke, which increases the risk of and complications from severe viral infections.
- Restrict your child’s exposure to groups of children during the cold season, particularly in day care centers.
- Immunize your child with SMA and siblings against the flu.¹

¹What is RSV and how can I protect my child from getting it? Parenting Corner, American Academy of Pediatrics web site: <http://www.aap.org>.

8 Perioperative Evaluations (Before and After Surgery)

Individuals with SMA are prone to certain complications after surgery, including respiratory infection and breathing problems. If these complications occur, your child may need to be intubated and placed on a ventilator for a period of time, and may even need a tracheostomy. As described in earlier sections, a tracheostomy calls for a small surgical hole in the neck, where a breathing tube (“the trach”) is inserted. The trach bypasses the mouth and vocal cords and goes directly into the large airway, known as the trachea. The best way to avoid complications is to evaluate respiratory health before and after surgery and take any needed precautions.



Evaluation before elective surgery (pre-operative) should include:

- Physical exam by a pulmonologist
- Measurement of respiratory function and cough strength
- Swallow evaluation, if warranted
- Chest x-ray
- Sleep study, if warranted

The care plan after surgery (post-operative) will depend on the results of the pre-operative evaluation and the type of surgery.

If there is pain with breathing or coughing after surgery, talk with the doctor immediately about pain medication for your child. Shallow breathing and a weak cough related to pain can lead to pneumonia.

If, after surgery, your child has severe problems with breathing (including sleep apnea), cough clearance, and poor muscle function, he or she may need to go into the intensive care unit.



What are Special Needs of Children with SMA Type I?

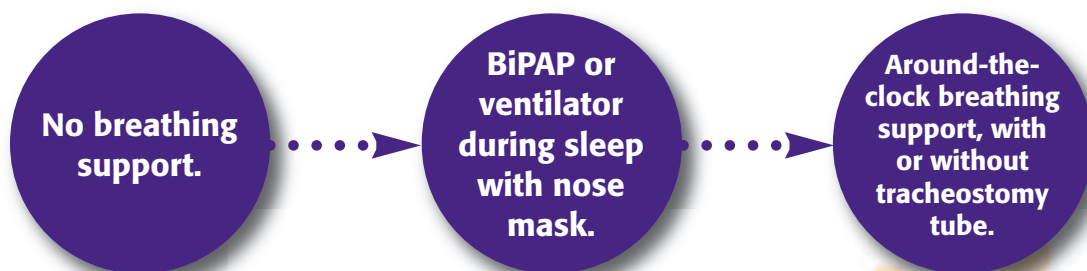
Children with SMA Type I face many physical challenges. There is no treatment to reverse or stop the progression of SMA Type I and there is no cure. There are, however, care interventions that will help with breathing and feeding problems, muscle weakness, bouts of illness, and discomfort.

Breathing Support

Each child with SMA Type I is unique and has a different set of issues and challenges in regard to chest muscle weakness and strength. Therefore, what your doctors, therapists, and you can do to help with breathing support varies from child to child. Their needs are difficult to predict.

The breathing support for children with SMA Type I can include a range of interventions even when they are well. During colds, most Type I children need extra breathing support. This may include BiPAP or a ventilator, either while asleep and possibly 24 hours per day, until the cold improves. Almost all children with SMA Type I will breathe better during sleep with BiPAP or a ventilator with a nasal mask, starting at an early age.

Type I Breathing Support



Respiratory Care Options and Difficult Decisions

Parents of children with SMA Type I face difficult decisions about respiratory care management. These decisions have to do with 1) non-invasive respiratory care, 2) invasive respiratory care, and 3) palliative care.

Ask your doctor to talk openly with you about what is involved in these three different approaches, including the advantages and disadvantages for your child, and what they mean for you as a caretaker. In these conversations, tell the doctors about your beliefs and values. What do you feel will add quality and meaning to your child's life?



Some parents are very aggressive about their child's medical care, seeking to extend life as long as possible. Others want a moderate level of medical care to minimize illness and promote strength and comfort. Others use very few interventions and allow nature to take its course. Finally, some parents use a combination of approaches. Loving, caring parents make different choices and there is no one right or best way.

What is best for one child may not be the best choice for another. Be sure to discuss all options with your doctors. Create a plan of action for different stages and points in the future, including times when your child's breathing situation changes. It is always better to have a plan and to think ahead, than to not have a plan and be caught off-guard in a crisis. You may need to act quickly in an emergency and will want to be as ready as possible. Remember, plans can always be changed and adjusted, based on your child's needs, and if you change your mind.

Non-Invasive Respiratory Care

You will be asked about *non-invasive* breathing support with a nose mask, or nose and mouth mask, for your child. The mask is attached to a BiPAP (Bi-level Positive Airway Pressure) machine or a mechanical ventilator, both of which deliver a continuous flow of air to the lungs. These methods and equipment are usually effective, but parents are advised to monitor closely with pulse oximetry. Many parents find that they need to be even more vigilant with their child's care during times of illness. See Section #7, Care During A Cold, for special steps to take with your child.

Many children tolerate the nose mask breathing support very well and can use it continuously. For a variety of reasons, however, the mask does not work well for every child or family. Common problems include skin irritation, pressure sores, poor fit, or the child just rejects the mask. The pulmonologist-respiratory therapist team can work with you to find a solution for your child. If your child has difficulty adapting to BiPAP, or if your child needs breathing support for 24 hours per day, you will need to make some hard choices about the next steps in your child's respiratory care plan.

Invasive Respiratory Care with Intubation

You will be asked about invasive approaches, including intubation, if your child is very sick, especially with a cold. Intubation will help your child breathe through a tube in the mouth. This tube can be used for a few weeks to a month or a tracheostomy (tube through small surgical incision in the neck) may be needed. Either tube is connected to a ventilator that supplies a continuous flow of air into the lungs. There is no face mask.

Remember, everyone becomes weaker with a cold. Your child may or may not recover from this cold or flu. Talk with your doctor to find out if the illness and breathing problems are temporary and reversible or a sign that your child is gradually becoming weaker overall.

If your child is intubated after surgery or because your child is acutely ill, it will be important for your child to receive aggressive airway clearance. This is extremely helpful while intubated and immediately after extubation (tube removal), when the child returns to non-invasive breathing support with a nose mask. The goal is to achieve a normal chest x-ray and to *not need* supplemental (extra) oxygen before extubation.



If your child is able to return to the mask, breathing support, and then does not tolerate it (despite the best efforts of the doctors and respiratory care therapists), you will be faced with these difficult decisions...should you...

- Seek a second opinion from doctors and therapists who are more experienced in non-invasive breathing support and SMA?
- Continue breathing support with intubation?
- Withdraw all interventions and let nature take its course?
- Transition to palliative care?

There is no one right answer and, as a parent, you can only do what you feel is best for your child and for your family. These are very personal decisions and they are different for each family.



Palliative Care

The word *palliative* means soothing, calming, and pain relieving. It is all about comfort.

There is no easy way to define the elements of palliative care for children with SMA. They are different for each child and family. You may decide that you no longer want to prolong your child's life if it includes more illness and suffering. But, you may want to help your child breathe more easily, with some degree of breathing support (such as BiPAP) and to be as comfortable as possible no matter what.

There are a number of gentle, non-invasive interventions that can help to keep your child very comfortable. Please discuss these with your doctors.

"The goal of palliative care is to add life to the child's years, not simply years to the child's life...to optimize the child's experience, rather than hasten death".²

"In the most severe cases of SMA, parents can be faced with anguishing decisions about therapies that they see as prolonging suffering rather than relieving the disease. Whenever possible, end-of-life care options need to be defined and discussed openly with parents and family members so that any decisions made reflect their values. It is important to have this discussion, however difficult, before a crisis occurs".³

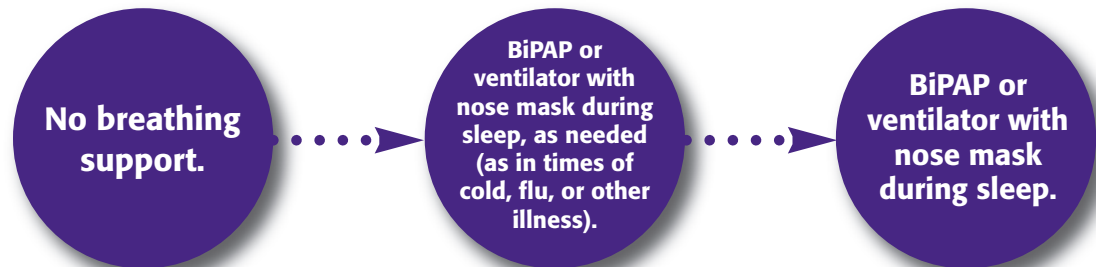
² From American Academy of Pediatrics, Policy Statement on Palliative Care for Children.

³ From the Consensus Statement for the Standard of Care in SMA.

What are Special Needs of Children with SMA Type II?

Children with SMA Type II rarely need a tracheostomy. In general, these children are strong and they can be managed with non-invasive respiratory care techniques on a day-to-day basis. This means that when the child is healthy, he or she can use BiPAP or a ventilator with a nose mask, or negative pressure ventilation during sleep; and the cough assist device, when needed. Some children with SMA Type II do not need any breathing support during sleep.

Type II Breathing Support



Care During A Cold

When children with SMA Type II develop colds, they may need to use the mask breathing support more often and sometimes continuously. They will be weaker than usual during the cold, but strength will return after the cold is over.

During the cold, they will need respiratory care treatments every 4 hours with airway secretion mobilization, cough machine use, and postural drainage. The cough machine may be needed more often to clear the mucus. See Section #7 on Care During A Cold.



What are Special Needs of Children with SMA Type III?

Children with SMA Type III are generally strong and have good respiratory function. In times of illness (colds, flu, infection), however, they may need non-invasive breathing support with manual cough assistance. Airway suctioning, chest physiotherapy, oximetry, and postural drainage should be considered, also as needed. Non-invasive breathing support, combined with airway clearance techniques, will reduce the need for intubation. Tracheostomies are not appropriate for Type IIIs.

In some cases, children and adults with SMA Type III can have breathing trouble that goes unrecognized, including obstructive sleep apnea and hypoventilation.

Routine assessments should include:

- Pulmonary function tests (spirometry, lung volumes, respiratory muscle function tests)
- Physical exam
- Measurement of cough effectiveness



Breathing Basics Summary

- Breathing and respiratory care problems are the leading cause of illness for children with SMA. They are the most common cause of death for children with SMA Type I and II.
- Respiratory care interventions (breathing support measures) are essential to the survival and comfort of children with SMA.
- Children with SMA Types I and II have very weak breathing muscles. This weakness makes it difficult for them to cough and clear lung secretions, and can cause poor breathing during sleep. Respiratory muscle weakness results in difficulty managing colds, flu, and other illnesses.
- Children with SMA breathe differently due to weak breathing muscles. The pull of their stronger diaphragm muscle causes stomach breathing, which results in a bell-shaped chest and sunken sternum bone.
- As a parent of a child with SMA, you will want to consult early with doctors, including a pediatric pulmonary specialist. Your child will need a personalized respiratory care plan with goals, including goals for times of illness.
- Children with SMA Type I face many physical challenges. Their breathing support needs vary and are difficult to predict. Breathing support can range from *no breathing support*; to *breathing machine at night with nose mask*; to *around-the-clock breathing support with or without a tracheostomy tube*.
- During colds, most Type I children will need extra breathing support, including a breathing machine for 24 hours per day, until the cold goes away.
- Parents of children with SMA Type I face difficult decisions about respiratory care management. These decisions relate to 3 key approaches to care: non-invasive respiratory care, invasive respiratory care, and palliative care. Parents should talk openly with the doctors about these different approaches, including advantages and disadvantages for the child and the family. Loving, caring parents make different choices and there is no right or best way.
- Children with SMA Type II rarely need tracheostomies. In general, these children are strong and they can be managed with non-invasive respiratory care techniques on a day-to-day basis. In times of cold or illness, they may need the nose mask and cough assistance during sleep.



- Children and adults with SMA Type III are generally strong and can be managed with non-invasive respiratory techniques during times of illness. They can have trouble with breathing that goes unrecognized, including obstructive sleep apnea and hypoventilation. Regular check-ups are important.

- Basic elements of a respiratory care management program:

1: Assessment, Evaluation, and Monitoring

Children with SMA need regular evaluations with the doctor every 3-6 months, generally, to assess breathing and look for any problems. Children with SMA Type III may need to be seen less often.

2: Breathing Exercises for Lung Development

Breathing exercises, usually done at home, can help children with SMA develop their lungs.

3: Coughing and Airway Clearance

Coughing and airway clearance are necessary to remove nasal secretions and inhaled particles. Helping children with SMA to cough more forcefully will help prevent lung infections and other respiratory complications. Non-invasive interventions include use of a cough machine and manual cough assist.

4: Removal of Mucus from the Airways

Children with SMA cannot cough strongly enough to remove secretions from their upper airways, especially during colds. Non-invasive techniques for removing secretions include chest physiotherapy (manual or mechanical percussion), postural drainage, intrapulmonary percussive ventilation (IPV), and high-frequency chest-wall oscillation or vest therapy.

5: Measuring Oxygen Levels

Children with SMA need extra breathing support when their oxygen saturation levels drop too low. A pulse oximeter is an easy, non-invasive tool for measuring oxygen level.

6: Breathing Support

There are both invasive and non-invasive interventions that help children with SMA to breathe more easily. Non-invasive respiratory care calls for an air-tight mask over the nose or nose and mouth. Invasive respiratory care calls for a breathing tube through the mouth or through a small surgical hole in the neck (a tracheostomy). In both non-invasive and invasive breathing support, the tubes are connected to a breathing machine that pumps air into the airways and lungs.

Children with SMA Type I and SMA Type II often need additional breathing support during sleep and during times of cold, flu, and illness.

7: Care During A Cold

During a cold, children with SMA may have difficulty clearing mucus from their lungs. Care interventions include airway secretion mobilization, use of cough machine, postural drainage, and pulse oximetry to measure oxygen saturation levels.

8: Perioperative Evaluations

Individuals with SMA are prone to certain complications after surgery, including respiratory infection and breathing problems. The best way to avoid these complications is to evaluate their respiratory health both before and after surgery and to take any needed precautions.

What is Families of SMA?

Families of SMA is a non-profit organization and the largest network of families, clinicians, and research scientists working together to advance SMA research, support families, and educate the public and professional community about SMA. Through numerous chapters in the U.S., and more than 55,000 supporters, FSMA raises millions of dollars every year for SMA research. This funding fosters basic and clinical research, drug discovery programs, and multi-center clinical trials. The ultimate goal of FSMA is to accelerate the discovery of an effective treatment and cure for SMA.



Every June, FSMA hosts the SMA Family and Professionals Conference. This brings together families, clinicians, and research scientists from around the world to discuss the latest breakthroughs in SMA research and care management. Parents and children find this a wonderful networking event and stimulating educational experience. *Please join us!*



Families of Spinal Muscular Atrophy

P.O. Box 196
Libertyville, IL 60048-0196



Phone: 1-800-886-1762
Fax: 847-367-7623



Email inquiries:
info@fsma.org



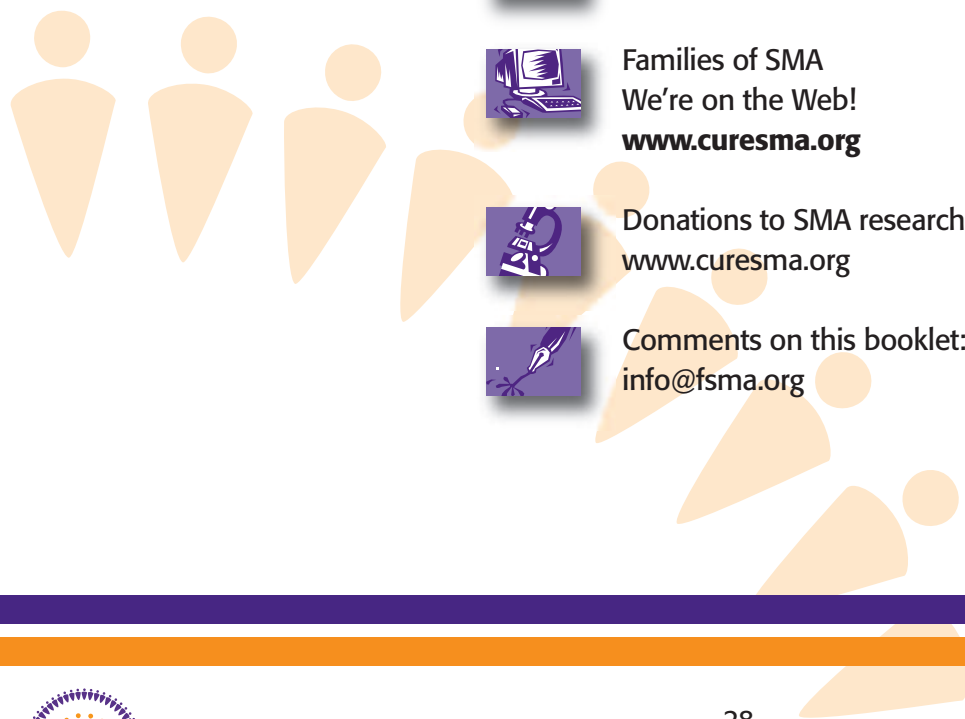
Families of SMA
We're on the Web!
www.curesma.org



Donations to SMA research:
www.curesma.org



Comments on this booklet:
info@fsma.org



Where to Go for More Information:

Families of SMA Website: www.curesma.org

Consensus Statement on Standard of Care in SMA:

<http://www.fsma.org/FSMACommunity/MedicalIssues/StandardofCare/>

Family Guide to Consensus Statement on Standard of Care in SMA:

<http://www.fsma.org/FSMACommunity/MedicalIssues/StandardofCare/>

What Respiratory Equipment Will You Need at Home?

Talk with your doctors and the respiratory care therapists about the equipment you will need at home. This may include the following:

- Cough machine
- Method for airway secretion (mucus) mobilization (manual or electric chest physiotherapy)
- Method for performing postural drainage
- Pulse oximeter to monitor oxygen saturation level
- BiPAP machine or mechanical ventilator
- Portable suction machine

Please note that oxygen tanks are typically not needed. However, some children with SMA Type I may require supplemental oxygen for emergencies.



For electronic copies:

Download this booklet from the FSMA web site at www.curesma.org. Go to the respiratory care section.

For print copies:

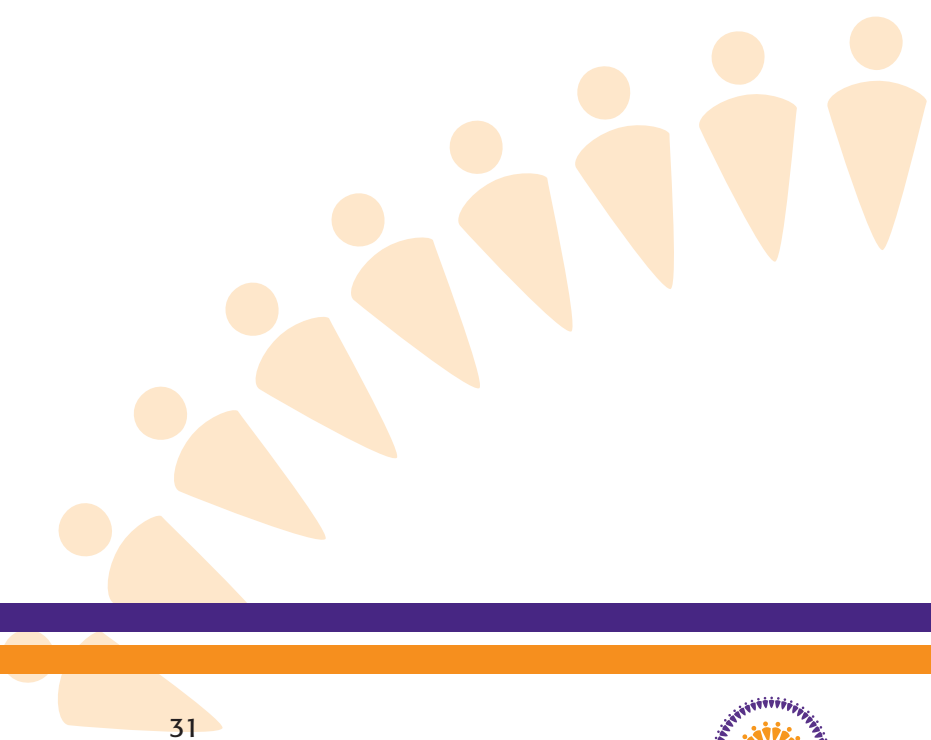
Please contact the FSMA national office at info@fsma.org.

Disclaimer:

Families of SMA does not, as an organization, support or endorse any particular treatment or therapy. Information contained in this booklet is for informational and educational purposes only. All medical information presented should be discussed with a qualified physician.

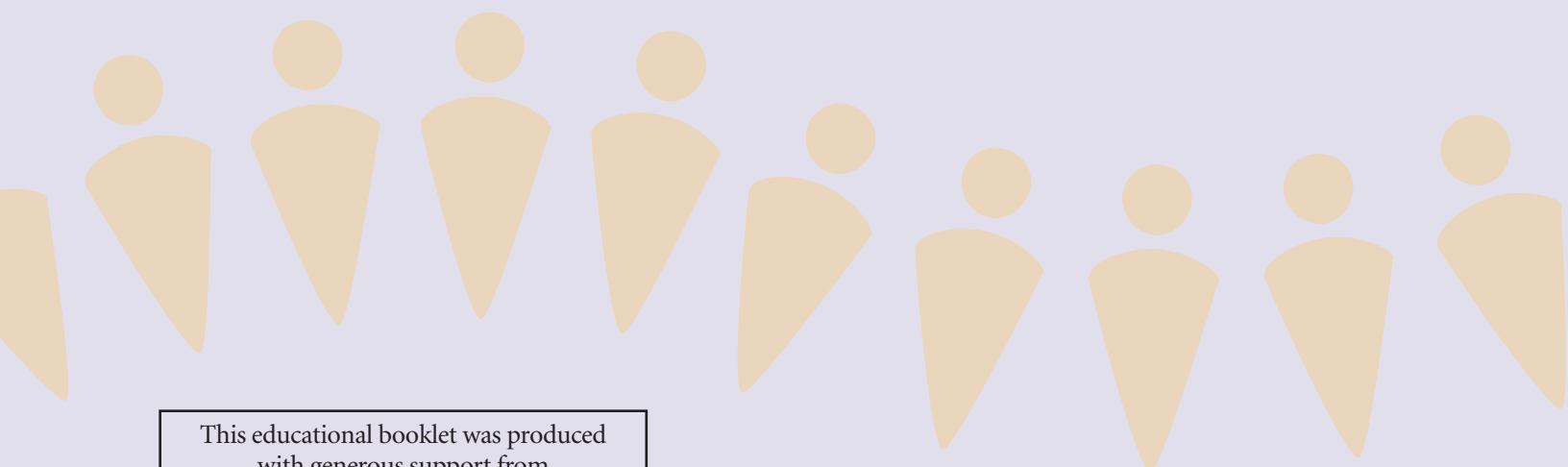
Date of publication: February 2009

©2009 Families of Spinal Muscular Atrophy





PO Box 196, Libertyville, IL 60048-0196
1.800.886.1762 • Fax 847.367.7623
www.curesma.org • info@fsma.org



This educational booklet was produced
with generous support from
The Angel Baby Foundation.