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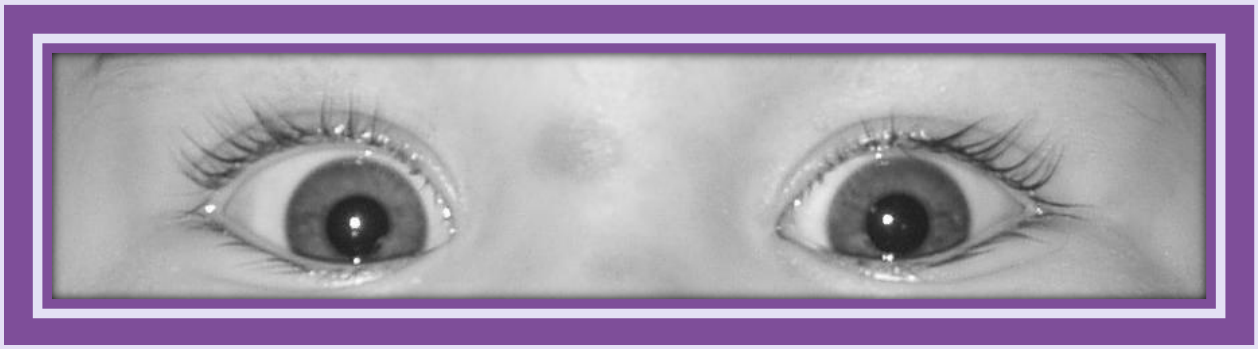


Deirdre Valeria's Daily Medical Care Handbook

Homecare for a Spinal Muscular Atrophy Diva

CREATED BY:
Vivian Rivera
&
Mark Medina

Contents

DEAR HOME CARE PROFESSIONAL:

Welcome to our home! Thank you for joining us here to take care of our precious girl, **Deirdre Valeria**, our beloved DeeVa (you may call her Deedee).

Deedee is our first and only child. She has always been a beautiful baby, so imagine our disbelief when she was diagnosed with Spinal Muscular Atrophy Type 1 at her 4 months of age. SMA Type 1 is the worst of SMA types, and most babies do not make it to their first year of life. The main cause of death is Respiratory Failure. So, if we seem a bit psycho about protecting Deedee from any sickness or allergy, please remember this fact. **She is very frail**, and although she may look as a normal toddler, looks can be deceiving.

Our DeeVa is a beautiful baby girl who looks strong and healthy, has charming gaze, grants a smile only if she trusts you, loves music and dancing lights. Even though she can't sit up or walk or eat by mouth, she still loves to have fun and play! **Please talk to her**, **sing to her and interact with her**. She enjoys it all, and responds to all. Deirdre is **VERY** sensitive.

We are blessed and thankful for each day with her smiles, joy, beauty and curiosity. We also are a bit worn out from her demanding medical needs. Some days are good and some are not. All are different, amazing… and draining.

We didn't expect to have a child with so many medical needs, so that also means we did not anticipate the need for nurses, therapists, doctors, or endless medical equipment that are important to Deedee's ongoing care. It can be quite tiring and stressful, draining on our family life, marriage, finances, etc. Because of this, we are VERY thankful and appreciative of the time and effort that you give to our little girl.

Please let us know if you have **ANY** concerns or questions. **ALWAYS** ask if you don't know the answer, if you are curious, or to make certain. Questions are **ALWAYS** good! We know almost anything that there is to know about the specific care that she requires. We have handled it all on our own from the beginning, learning through other parents who supported us as they face the same challenges. They keep supporting us through the SMA Support Chat, where we meet daily and

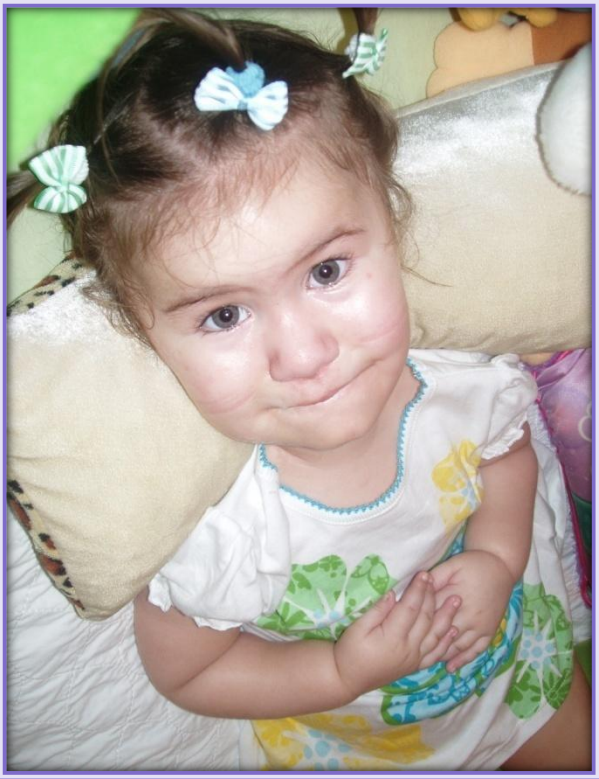
talk about our concerns and battles. We also have the support of the three doctors who are the experts about SMA children: Dr. Bach (UMDNJ), Dr. Swoboda (Utah) and Dr. Schroth (Wisconsin).

Whatever information we share with you comes from a **reliable source** (the specialist doctors, Families of SMA organization, books of Neurology focused on SMA, and the official websites within the University of Utah's Neurology Department, and within UMDNJ Non-Invasive Respiratory Clinic Page). We have compiled here all relevant information, nothing is made-up.

SMA is a **rare disease**, with **NO CURE**. All we can do is fight it with our hearts set on granting our DeeVa everything she needs for survival, everything she needs for growth, and everything she needs to be someone of worth in our society. All of this we believe is possible. We see Deedee's needs as CHALLENGES not problems. We want her to have the world at her hands.

Deirdre has **NO EXPIRATION DATE**. We expect that you internalize that fact. She is very intelligent and demanding. We don't know how long she will stay with us, but we believe in living each day to the fullest, and having ongoing growth (especially spiritual growth). God granted us our wish, allowing an angel to dwell among us. She is a miracle, a gift of love. Let us help her fight for her life, for her future.

Vivian Rivera & Mark Medina



YOU CAN REACH US ANYWHERE, ANYTIME. OUR BABY IS OUR **FIRST PRIORITY** ALL THE TIME, SO IF YOU HAVE TO CALL US, PLEASE DO SO. THIS APPLIES DAY OR NIGHT. WE HAVE VOICE MAIL IN OUR PHONES AND WE CHECK OUR EMAIL DAILY.

Home Phone: (973) 878-3434
Vivian Rivera:(973) 517-5909
Mark Medina :(973) 517-7407

WE ENCOURAGE YOU TO GO VISIT OUR BABY's INTERNET WEBPAGE AS IT HAS LINKS TO SMA INFORMATIONAL SITES. THE MORE YOU LEARN ABOUT THIS DISEASE, THE BETTER CARE YOU CAN PROVIDE OUR "DEEVA".
Deedee's website, and diary is found at: www.alittlemageinthefamily.blogspot.com
Also, you may visit her fundraising web page: http://www.geocities.com/deirdremedina@ymail.com

ABOUT MOMMY

Mommy is the serious one. She is artsy and creative (painting, drawing, scrapbooking, blogging, web-designing). She has a BSBA in Computerized Information Systems and Marketing, and a MBA in Human Resources and Marketing. She worked 9 years in Vocational Rehabilitation Administration of Puerto Rico as Systems Administrator, and has 11 years of experience in the Information Technology field. She is currently completing a Webdesign Diploma at home while taking care of Deedee.

Vivian Rivera:(973) 517-5909

Email: v_rivera_pr@yahoo.com

ABOUT DADDY

Daddy has a wacky personality, but don't be fooled… He is CNA (Nurse Assistant), has an Associates Degree in Computerized information Systems and has an almost complete BBA in Computers. He worked for 5 years in Cordis, Johnson & Johnson in Puerto Rico as production operator. A loving father, he has a 5-year-old daughter, Amanda, besides Deedee. He wants to become an RN so he can take care of Deedee all the time while getting a stable pay.

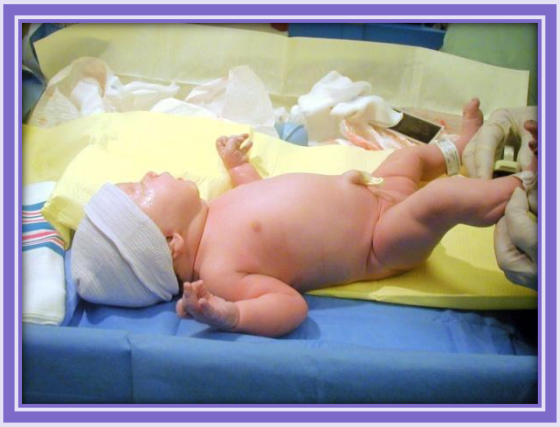
Mark Medina :(973) 517-7407

Email: mark_medina_pr@yahoo.com

GENERAL GUIDELINES FOR CAREGIVERS

- **BE ON TIME**. Please call if you are running late, or if you have a special situation.
- **WASH YOUR HAND BEFORE TOUCHING DEEDEE**. You should use sanitizer at all times. Use gloves when doing deep suctioning and respiratory treatment.
- **NO PHONE CALLS**. If you must make a call, ask us so you may use our home phone. You may receive phone calls from your company on our home phone. Remember you are here to give a professional service.
- **DO NOT FALL ASLEEP**. If you are tired or feeling ill, go home and rest. Deedee's safety and wellbeing requires complete awareness. An emergency happens in seconds.
- **INTERACT WITH DEEDEE**. She is smart, sensitive, and notices EVERYTHING. Give her choices and emphasize names, colors and shapes so she learns and increases her skills. Just because she cannot move it doesn't mean she is a vegetable!
- **FILL YOUR PAPERS AS WELL AS OUR NOTEBOOK**. We keep a personal record of oxygenation/heart rate/ diaper change/ temperature by the hour, so we can track problems.
- **IF YOU ARE SICK, DO NOT SHOW UP**. If someone around you is sick, the same applies (other case, anyone in your family). This is to avoid CROSS-INFECTION. A simple cold can KILL our girl, so this rule is VERY important and is enforced with NO EXCEPTIONS. Colds turn into pneumonia within hours and are the main cause of death in SMA infants/toddlers.
- **WE BELIEVE IN TEAMWORK**. Taking care of Deedee may look simple, but it can get very complicated if the schedules are not kept. The best care is between 2 people. Remember that one of us (mom or dad) is always in the house to help take care of her, as we know how things can get out of hand. We have been there at Deirdre's worst moments, and we know what to do in any circumstance, so be certain that you are not alone.
- **COMMUNICATION IS KEY**. If you have doubts, ASK! If you have ideas and/or concerns, let us know. Although we keep ourselves updated with everything SMA related, you may know something about the "broader scope" that we do not know because of our specific focus.
- **A NOTE ON OXYGEN:** Oxigen should not be used unless Deedee has respiratory failure or is in high distress (numbers stay within 70-80 for long after suction and ambu is used). Why? 90% of the time numbers go down because of a mucus plug on her throat. Always suction, them ambu, then use CoughAssist. Always call mom or dad when that happens in case something bigger happens.

ONCE UPON A TIME: DEEDEE's STORY



Deirdre Valeria was born on **AUGUST 17, 2007** in San Germán, Puerto Rico via C-section. A lovely newborn, we all were bewitched by Deedee's peaceful demeanor.

Deedee's first big party was her **Baptism** on October 21st, 2008. The event took place at our favorite church, Fatima, and afterwards we did a small gathering at baby's great-great aunt's home. Balloons, cake, music… She had it all. And she looked gorgeous with her long white attire.

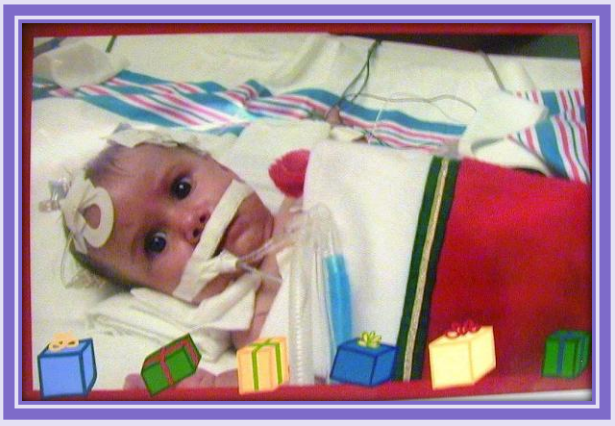
Deedee's first **Halloween** arrived, and after some debate and realizing it was really hot, she dressed up as a devilette, just like her mom. Her dad dressed up as pirate and her godfather dressed up as zombie. It was really fun!

For her first **Thanksgiving** we wanted to begin some family traditions and we decided we would invite both our families to our home to celebrate… So, we had turkey, a nice dinner, guests from both sides of the family, and a nice picture session.

Deedee's first **Christmas** turned out to be… unforgettable to say the least. We were preparing everything for a Christmas night gathering at home, but things took a wrong turn a week before the holiday.

On December 17th, Deedee had a respiratory arrest. She spent one month in intensive care at San Antonio's Hospital in Mayaguez. Both her lungs collapsed. Doctors didn't give us much hope on her survival. On Christmas day all we asked for was for the gift of life for our precious 4 month old baby. God granted that.

From the moment that Deedee went into PICU, doctors speculated she had SMA and so we started an internet search for what that meant and the choices available for treatment. Upon my mentioning the disease on the baby's online diary, I began to receive emails with information about the illness and the best doctors/experts to treat it. Parents from US and Canada sent us in the right direction towards helping Deedee. We got in touch with SMA expert doctors Dr. Bach, Dr. Swoboda and Dr. Schroth, each of whom lead us



towards what to do while trying to coach doctors in the island (which was really hard as they did not want the coaching). Deedee was **diagnosed** with the fatal Spinal Muscular Atrophy on **January 3, 2008** after the hospital received the tests sent by their geneticist.

We became experts on the immediate care for this illness. We were advised not to allow tracheotomy as in most cases that was not the right solution for babies with SMA. The hospital did not take it lightly when we started telling them what to do. In **January 8th** Deedee was transferred to the Pediatric Hospital in San Juan, Puerto Rico mainly because the hometown hospital had reached its limit on knowledge of what to do.

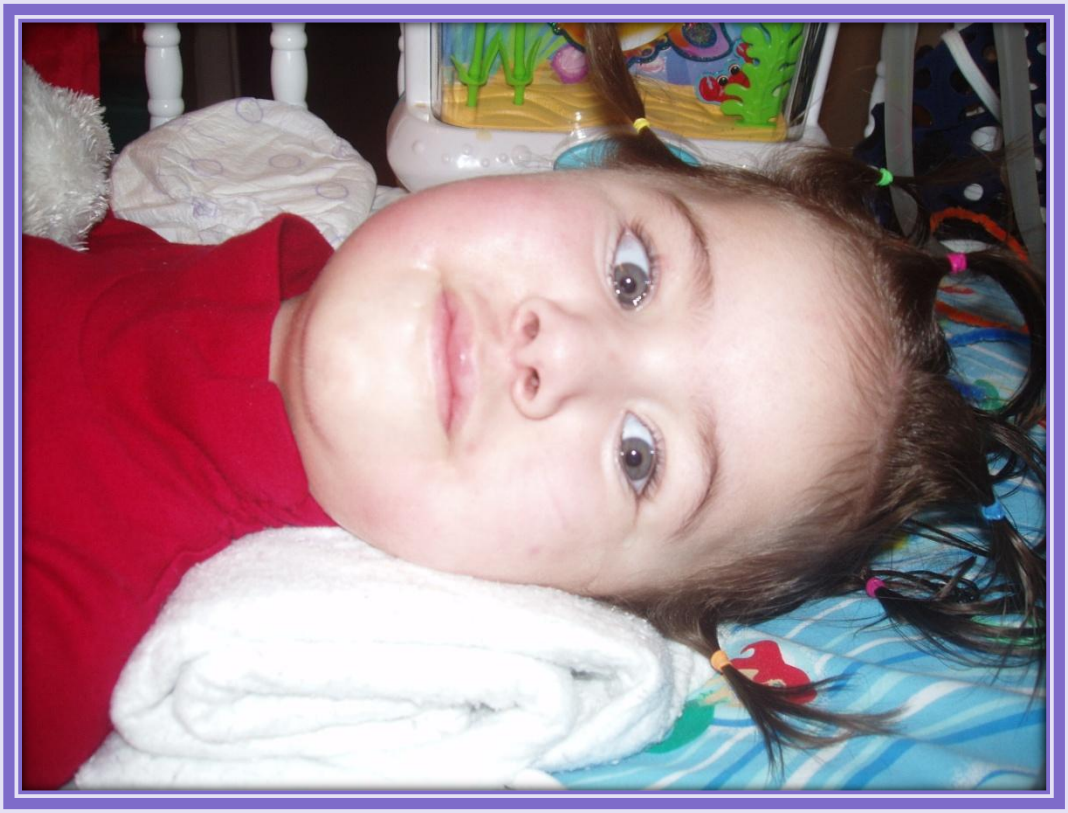
At San Juan we were received by Dr.Diaz, a doctor that was open to the idea of Non-Invasive treatment and who tried to apply it over there. The big problem was that since they had not done it before, it was all trial and error. We feared that so many inconsistencies from therapists and the different doctors' approaches (as shifts change) would not be favorable for our baby. They performed the **G-tube** operation adding the **fundoplication** at our request (we had been advised by Dr. Swoboda to do it to prevent reflux). They wanted to trach Deedee but we were opposed to that. Research and observation of Deedee's behavior let us know she would make it without a tracheotomy, if given the right chance.

What followed were long days of constant fights and bad news regarding services and people not doing what they were supposed to… It was nerve-breaking as in PR you are not allowed to stay all the time with your baby. We had a special permission to visit from 10am to 9 pm. We made sure that while we were there things were done right, but we feared (with good reason) that things were not so by the book when we were not there at night. It was all very stressful, handling the meetings to get help, plus the hospital, plus the fight with the agencies that were supposed to help but didn't want to… Nevertheless, at all moments God was there, and whenever a door closed He opened a window.

Two months later, after a **fundraiser** led by Deedee's godmother, and after finally getting the money for an **air ambulance** from Catastrophic Funds, Deedee was transferred in **March 28th** to University Hospital at Newark, NJ. Even the flight was troublesome as Deedee got in a shock ever since she was taken from the hospital room… until she was placed in the new room at Newark. She didn't even blink throughout the flight. The nurse suggested that I carried her in my arms. I carried precious

Deirdre for the first time after more than three months.

Deedee was extubated on the third day of her arrival, and then stayed in intensive care for two more weeks. On **April 14th** she was finally allowed to go home following the Non Invasive treatment that made her Bipap ventilator dependent (and that prevented her from being trached, for now). **We decided to stay in Newark to be able to give our baby the best medical treatment for her condition.**



It all has been hard on us as we are alone in a new place. We know no one. It is basically starting a life from scratch. We are focused on getting Deedee's care, and on getting a job so we can be back to normal. Our family and friends are in Puerto Rico, which makes everything even harder. But, we have the support from the **FSMA** parents, our new big family, who have guided us all along. Without them we really don't know what would have happened, or even if Deedee would be alive today. So far, everything has been very positive for Deedee. Her therapists from Early Intervention visit her regularly. Prevention is key for SMA kids, and so she gets at home respiratory treatments from her mom and dad twice a day. We hope that your help will give us some relief to improve even further Deedee's daily medical care and quality of life.

WHAT IS SMA?

Spinal muscular atrophy (SMA) is a **genetic, motor neuron disease** caused by progressive **degeneration** of motor neurons in the spinal cord. The disorder causes weakness and wasting of the voluntary muscles. Weakness is often more severe in the legs than arms. The childhood SMA's are all autosomal recessive diseases. This means that they run in families and more than one case is likely to occur in siblings or cousins of the same generation. Parents usually have no symptoms, but still carry the gene. The gene for SMA has been identified and accurate diagnostic tests exist. There are many types of SMA. **Deirdre has Type I SMA**.

SMA type I, also called Warding-Hoffmann disease, is evident before birth or within the first few months of life. Symptoms include floppiness of the limbs and trunk, feeble movements of the arms and legs, swallowing and feeding difficulties, and impaired breathing. Affected children never sit or stand and usually die before the age of 2.

Symptoms of SMA type II usually begin between 3 and 15 months of age. Children may have respiratory problems, floppy limbs, decreased or absent deep tendon reflexes, and twitching of arm, leg, or tongue muscles. These children may learn to sit but will never be able to stand or walk. Life expectancy varies.

Symptoms of SMA type III (Kugelberg-Welander disease) appear between 2 and 17 years of age, and include abnormal manner of walking; difficulty running, climbing steps, or rising from a chair; and slight tremor of the fingers.

Kennedy syndrome or progressive spinobulbar muscular atrophy may occur between 15 and 60 years of age. Features of this type may include weakness of muscles in the tongue and face, difficulty swallowing, speech impairment, and excessive development of the mammary glands in males. The course of the disorder is usually slowly progressive. Kennedy syndrome is an X-linked recessive disorder, which means that women carry the gene, but the disorder only occurs in men. Congenital SMA with arthrogryposis (persistent contracture of joints with fixed abnormal posture of the limb) is a rare disorder. Manifestations include severe contractures, curvature of the spine, chest deformity, respiratory problems, an unusually small jaw, and drooping upper eyelids.

Is there any treatment? What is the prognosis? **SMA has NO cure**. Research is ongoing, and everyday we get closer to a cure. Treatment of all forms of SMA is **symptomatic and supportive** and includes treating pneumonia, curvature of the spine, and respiratory infections when present. In addition, physical therapy, supports, and rehabilitation are useful.

The prognosis for individuals with SMA varies depending on the type of SMA and the degree of respiratory function. The patient's condition tends to deteriorate over time, depending on the severity of the symptoms.

What research is being done? The NINDS supports research to study gene function in SMA. Researchers have found the specific gene that, when mutated, causes SMA. Several animal models of the disease have been developed as well as tests that can determine SMA gene function. This allows scientists to screen drugs that may be useful in treating SMA. The NINDS has established a model translational research program to accelerate the process of developing a safe and effective treatment for SMA. More information about this program is available at **http://www.smaproject.org/.**

SMA is a scary disease and we know Deedee's prognosis but we also have a lot of hope for Deedee. She has overcome so much in her life already, so we are hopeful that she will exceed the prognosis. There are many children who have a great quality of life with SMA Type I and are older than 2 years old!In fact we look up to MJ who is SMA Type1, is 21 years old, and is completing her BS in Rehabilitation Counseling.

SMA is the LEADING genetic killer of children under the age of two. The National Institute of Neurological Disease and Stroke (NINDS) says that out of the 600 disorders it tracks, SMA is one of the closest to being cured. In one of the blogs, it talked about all the parents who were in Washington last year and will be there again on April 22nd lobbying for the proposed SMA Treatment Acceleration Act. The legislation would establish a national clinical trials network to develop safe and effective treatments for SMA patients and possibly lead to a cure for this disorder which strikes as many as one in 6,000-10,000 infants. "It's so close," the author of the blog said. "If we had parity in funding from government, it would be a done deal."

For more information about SMA visit: www.fsma.org, www.smasupport.com, www.doctorbach.com, www.fightsma.org and Deedee's online diary and website: www.alittlemageinthefamily.blogspot.com

Please make yourself acquainted with the CONSENSUS OF CARE FOR SMA PATIENTS included in this booklet's apendix. It was published by MDA in November 2007 and gives an overview of what should be the care, treatment and outlook about SMA.

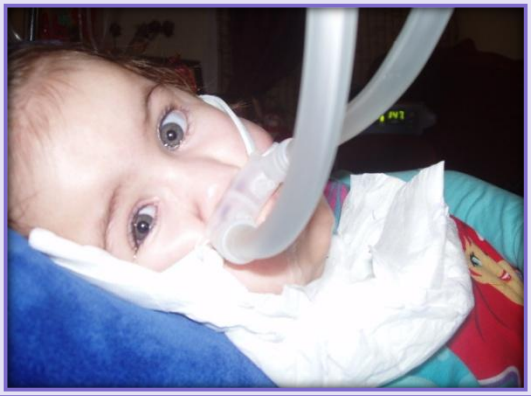
In October 2008, SMA was listed within a new Social Security Law as an illness that will have its immediate attention.

Right now, there is a PETITION TO CURE SMA. Help cure the #1 genetic killer of children under 2! The target is the US United States Senate and United States House of Representatives. Please sign the petition so projects of research to find the cure for SMA are sponsored by the government. http://www.petitiontocuresma.com/

DEIRDRE's SMA NON-INVASIVE VENTILATION PROTOCOL

When you are told that your child has SMA, most physicians give you one of two routes to consider:

- Palliative care by "letting them go" if they are in respiratory distress
- Ventilation via a tracheotomy
- NON_INVASIVE ventilation. Some of the leading experts in SMA (Dr. Schroth, Dr. Bach, and Dr. Swoboda) feel that non-invasive ventilation is an excellent option to help facilitate breathing and coughing. (For more information see Dr. Swoboda & Dr. Schroth CHOICES in the Appendix).



DEEDEE's care follows the NON-INVASIVE PROTOCOL. This involves **monitoring oxygen saturations and using devices to assist the child when necessary**. Air is administered through a bipap which gives intermittent positive pressure ventilation. T**his is NOT oxygen. DO NOT use oxygen unless absolutely necessary.** The ONLY time Deedee may require supplemental oxygen is during an emergency when she might have a "plug" that needs to be cleared, when she is ill, or upon respiratory arrest.

ABOUT OXIGEN:
Oxygen is in our home in Deedee's room. We also have oxygen near the ambu bag hanging from the feeding pump rack in her room. See O2 warning and remember that SMA kids should not be given too much O2 and rather the smallest amount of O2 needed to keep their saturations up and ONLY when necessary. If you feel Deedee may need 02 please ask Vivian or Mark first, UNLESS it is an emergency situation. Give 2L.

One of the most common treatment errors for SMA kids is oxygen therapy which expert Dr. Bach says is like "putting a band-aid on cancer". Oxygen turns off the brain's drive to breathe and increases the likelihood of ventilatory failure, allowing the blood's CO2 to increase to levels that render someone unconscious and cause them to stop breathing. Usually decreased oxygen saturations are caused by airway secretions that Deedee cannot clear on her own. Please use the respiratory protocol and do not use TOO MUCH O2 but rather only the amount needed WHEN it is needed. If you feel she needs any 02 please discuss with Vivian or Mark, unless it is an emergency.

THE EQUIPMENT

Deedee uses five main pieces of equipment every day. They are listed below. If you are not familiar with this equipment or have any questions please let me know. Remember – ASK if you are unsure about anything.

LTV 950 – We use the LTV 950 non-invasively as a Bipap. Deedee uses this during naps and night-time sleep. We use a humidifier at night, attached to the LTV tubing.

Her current settings are:

- ♥ Breath 24,
- ♥ Tidal Volume 170,
- ♥ Pressure Control 18,
- ♥ Inspiration Time 1

Please DO NOT change her settings at any time. Vivian or Mark may periodically change her settings based on her pulmonologist orders. If you have any questions about these please let us know.

Cough Assist – The cough assist helps Deedee cough and clear her airway. Since SMA kids have a very weak cough this is an essential part of her respiratory therapy. She uses this twice a day during her treatments or as needed. Her settings are:

- ♥ 40/40 (should NEVER go above this for any reason)
- ♥ Inhale: 2 seconds
- ♥ Exhale: 2 second
- ♥ Pause: 1 seconds
- ♥ Please DO NOT change these settings, and please check them every time you use the Cough Assist as the buttons turn easily.

If Deedee's 02 saturation ever drops to 94 or below we will cough and suction her to try to correct the problem. We also use the cough assist if she is having trouble clearing her throat and may need a little extra help. Please read her daily therapy section for more information. CoughAssist will help keep her lungs clear and help avoid major plugs that can threaten her life. If you can hear secretions in Deedee's lungs, the CoughAssist (preceded/followed by CPT) is the best way to bring those secretions to the surface so you can suction them out.

Pulse Oximeter – We monitor Deedee's oxygen when she is sleeping or any time we feel she may be low. Her oxygen may be monitored or "spot-checked" throughout the day as well. You can use it around the house, its battery lasts up to 8 hours without electric cord. Normal stats for Deedee are:

- Awake, with the BiPap: 98-100 /120-150HR
- Awake, without the Bipap:96-100 /130-160HR
- Sleep with the BiPap: 98-100 /90-125HR
- Sleep without the Bipap: ALWAYS HAVE BIPAP ON WHILE SLEEPING OR NAPPING TO PREVENT APNEA

Suction Machine – The suction machine is used as needed during Deedee's treatments as well as any time during the day she has a lot of secretions and needs extra help getting them out. Unless we feel Deedee absolutely needs it we try not to deep suction her all the time – this can cause irritation in her airway. We use a Yankauer (wand) to suction out her mouth, and a red catheter #10 for suctioning the nose. If Deedee is ill or has a lot of heavy, thick secretions she may require deep suctioning more than twice a day.

Feeding Pump – Deedee uses the Kangaroo Joey Feeding Pump all the times. Remember she cannot be without food for more than 4 hours, she needs the energy. The feeding bags cannot be removed for cleaning after being placed on the pump, or they will give error afterwards (leading to changing to a new bag). To clean the bag, simply pour water on the bag and prime it. Feeding is set to 200ml at 135/hr when feeding every 4 hours. At night, we give a double feed that is continuous for almost 6 hours: 400ml at 70/hr.

DAILY ROUTINE: THE FIRST SHIFT

(8am to 4pm)

8:00am	Change feeding bag, flush g-tube. Give vitamins (1ml) through G-tube. Feed #1 If baby is awake: Do not take mask off until she gets her respiratory treatment! Wipe her face & drool, put Vaseline in baby's lips. Comb her and change diaper. Change baby's position Turn on DVD with Baby Einstein Do Range of Motion exercises while baby watches DVD
9:00am	Play practicing grab/let go. Play with switch toys.
10:00am	Respiratory Treatment #1 Vibrations (each side for 5 minutes), suction as needed using saline bullets, and Cough Assist 3 times of 5 repetitions Oral care. Wipe baby's face. Apply Aquaphor around mouth-cheeks area and in lips. Place in wagon or stroller and take out to the living room.
11:00am	Someone from Early Intervention may visit (PT, OT)
12:00pm	Feed#2 Baby may usually nap at this time. If not, she will do so after her bath.
1:00pm	
2:00pm	Bath in tub using bath chair. Place pulse-ox sensor in ear before getting wet. Range of Motion exercises, play, speech therapy, and then bath. If washing hair, it should be the last thing done. Take baby into bedroom, make sure she is well covered by towels.
3:00pm	Massage with lotion. Put Vaseline in buttocks and rash cream in front. Pour some baby powder before closing diaper. Dress-up Place in wagon or stroller and take out to the living room. Comb baby.

DAILY ROUTINE: THE SECOND SHIFT

(4pm to 12am)

4:00pm	Feed #3 Place 6oz Miralax on the feed. Play movie on TV
5:00pm	If baby falls asleep to nap, make sure that you put on her Bipap mask to prevent apnea and give her lungs a break.
6:00pm	Flash cards
7:00pm	Play movie Range of Motion exercises while she watches it
8:00pm	Feed#4 Do Speech Therapy exercises
9:00pm	Take to bedroom Show flash cards to practice naming and colors Read a book Adapted/switch toy play
10:00pm	Respiratory Treatment #2
11:00pm	Prepare for sleep After treatment baby is placed on the side

DAILY ROUTINE: THE THIRD SHIFT

(12am to 8am)

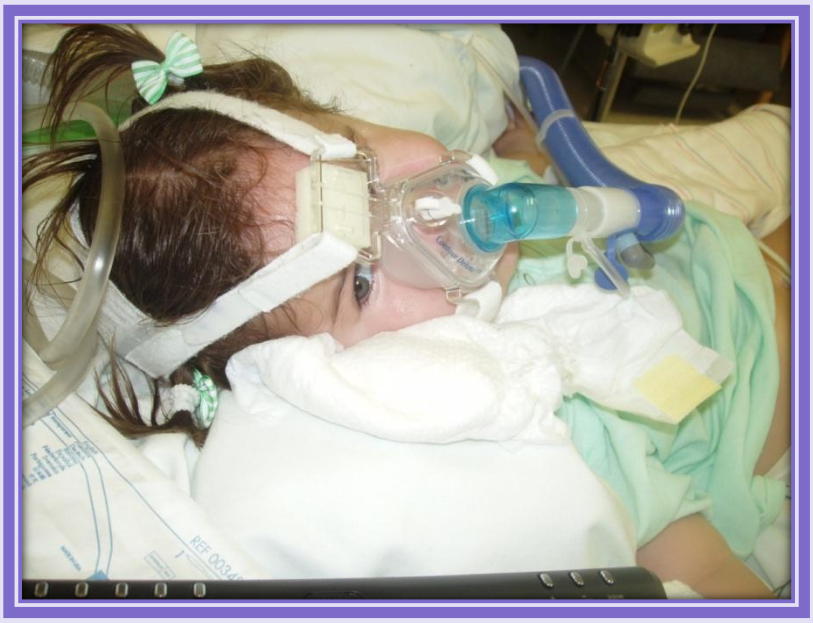
12:00am	Feed #5&6 in one, setup as continuous feed. 400ml@70hr, should end up around 5:30am Under normal circumstances, baby sleeps through the whole night without problems. When the pulse-ox beeps continuously, it is usually because the sensor is not reading. Baby slightly moves her legs, feet, and toes, so you may have to play with the sensor's location. We find that taping it to the leg side is better than taping it to the foot, it shows stable numbers usually 98-100. If it is evident that baby is in distress, we repeat the respiratory treatment (chest pt, vibrations and cough-a-lation as needed).
4:00am	Check baby to make sure everything is fine

Always keep these in mind:

- If at any moment Deedee looks distressed, put on the mask. Also, please suction anytime Deedee is coughing (sounds as choking) or has excessive secretions in her mouth.
- If she continues trying to coughing or sounds choked after suctioning in her mouth, please use manual AMBU if not in her room. If Deedee is in the crib, Cough Assist right away.
- While watching TV Deedee likes to have something in her hands (wand, spike-gel ball, switch for an adapted toy).
- Put on her leg splints, leave for 4 hours.
- If she falls asleep (nap) place the mask right away.
- Try to do face therapy with toothbrush vibrator (passing cheeks, surrounding mouth) each hour if possible.
- During Deedee's bi-pap break/naptime, this is a good time to have your lunch, make any necessary phone calls, paperwork, read a book (anything but falling asleep).
- Please also make sure to clean her medical equipment area, wiping with alcohol pads, cleaning out suction machines and tubing, refilling suction and "flush" water, refill diaper bin/wipes (all once per shift)

Thanks for taking good care of Deedee! We appreciate your efforts and care.

DAILY RESPIRATORY TREATMENTS



Generally Deedee's respiratory treatments are done twice a day – every 12 hours, one in the morning and one in the evening – unless she is ill. When she is ill we will increase them to every three to four hours around the clock, or as needed.

The following is a description of her treatments. Deedee likes to listen to one of her CD's (vivid music) while having her treatment. If you have any questions about this at any time, please NEVER hesitate to ask.

STEP 1: PERCUSSIONS to move secretions

- Chest Vibrations: Use the electric vibrator to do this CPT session. CPT should be done for 5 minutes on each quadrant (front right, front left, back right, back left) – 20 minutes total.
- When doing front right and front left pay special attention to vibrating the apice area. Only do CPT where she has lungs. Sometimes Deedee may need to be suctioned during or after her CPT.
- Trendelenburg positional drainage: This is "feet elevated above her chest" position. Do this with changing settings of position bed. Put her in this position and leave her for 10-15 minutes so secretions go from lower area of lungs to upper lungs. Sometimes Deedee may need to be suctioned during or after this.
- Chest Percussions (CPT): We prefer to do it using our hands. To get more control and target specific areas (such as apices). CPT should be done counting 50 times each quadrant (front right, front left, back right, back

left). Only do CPT where she has lungs. Sometimes Deedee may need to be suctioned during or after her CPT.

Step 2: COUGH ASSIST

Settings are set at 40/40 and should NEVER go above this. Inhale is set at 2 seconds, exhale at 2 seconds, pause 1 second. Always check the settings every time you use the Cough Assist as the buttons can turn easily.

- Deedee must be laid FLAT while she is being coughed, with a pillow under her neck to keep her airway open. We use a specific roll/pillow for this . Make sure to ask for it, so she is well supported.
- Wash your hands or use hand sanitizer before starting. Use gloves.
- Suction Deedee through nose and mouth using a few drops of saline solution and a #10 red rubber catheter.
- Deedee receives 3 sets of 5 coughs (Suction Deedee through nose and mouth using a few drops of saline solution and a 10 red rubber catheter in between sets).
- Turn the machine to automatic and the coughs will start. Verify the numbers are at 40/40, 2-2-1.
- Place the mask over Deedee's face and make a seal by placing hand in C position over mask, while holding her forehead with your other hand to tilt her head a little, opening her airway
- Air will push in (inhale), then push out (exhale) and pause. Count 5 repetitions (watch that her chest rises and goes down 5 times), and remove mask from Deedee's face placing the machine back on manual during the break.
- It is important to suction Deedee through nose and mouth using a few drops of saline solution and a #10 red rubber catheter in between sets.
- If you must do another coughing set, it is important to wait at least 30-45 seconds in between each cough set. If she is fussy you may do the whole session while the DVD is on, so her attention shifts and she calms down.

Step 3: ORAL HYGIENE

Once all the respiratory treatment is finished please wipe Deedee's face with a baby wipe. Use a toothette dipped in water and wipe inside Deedee's mouth and gums. Suction out as needed. Follow-up with putting Vaseline or Aquaphor on her lips with a Q-tip.

WHEN TO USE THE BIPAP

Use during naps, nighttime, daybreaks, and more often when she is sick. This machine gives Deedee a rest so she can sleep well during her nap/overnight while giving her lungs a break. SMA kids run a marathon each day because they work so hard to breathe, but the bipap gives them the break they need.

Deedee will need her bi-pap during naps and overnight. Typical naps are: After midday AND late afternoon for an hour or two. This all depends on when she wakes up and what is happening that day. In a normal day, Deedee can stay up to 9 hours without using the bipap on daytime.

Setting up BIPAP:

- Place her flat in her bed with either hand.
- Start the machine BEFORE placing the mask on Deedee's face.
- Once the machine has started place the mask squarely over Deedee's nose. If it doesn't seem to be squarely on her face or is in her eyes, start over.
- If Deedee's eyes are watering, it may mean there is a leak or the mask is too close to her eyes and causing her discomfort. ALWAYS TURN MACHINES ON BEFORE PLACING ON DEEDEE.
- Do NOT tighten mask too much or it will cause sores and Deedee will cry. It does NOT have to be too tight to have an effective seal. However, there must be a good seal in order for the machine to work properly.
- Once it sounds like a good seal, reposition Deedee on her side. We alternate sides each time she goes down for a nap. Use pillows, rolled towels, rolled blankets, and stuffed animals to put her in a comfortable position on her side. Place a blanket between her legs for support. Do not be alarmed if she has some secretions/drooling when using the nasal mask.
- Put a small diaper (size 1) between her face cheek and the pillow. We call it the "baba collector" as it keeps all drool from wetting pillows or her clothing
- She fusses a little until she is entertained, or until she falls asleep. Her heart rate will continue to go down when she is going to sleep and may stay between 90–120 while sleeping. Sometimes it is lower and sometimes a little higher.
- If stats are stable after her morning respiratory treatment, don't put the bipap right away. Deedee can stay for a few hours off the bipap, it all depends on how she feels. When she is off bipap, she needs CONSTANT supervision as it is when she can be more at risk of choking.
- When saturation drops to 95, suction and if she stays in 95 put her back on bipap.
- If baby looks distressed, seems tired, face flushes, cheeks are reddish and she seems fussy, put the bipap on.

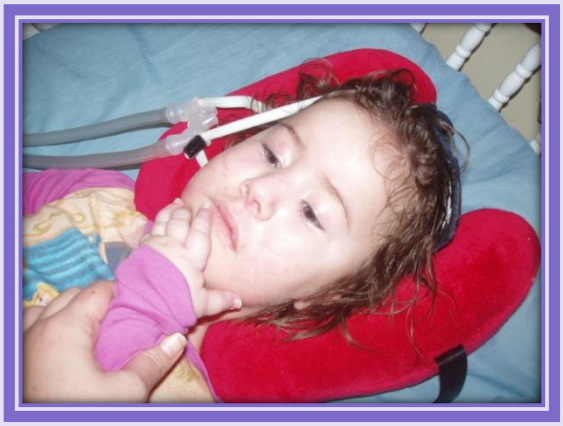
WHEN TO REMOVE THE BIPAP:

When Deedee's numbers are constant and high, we allow her to be without the bipap. Normally she can be up to 9 hours without the bipap, under normal conditions.

- If you have just placed the bipap on her face and she is continually crying, check for any leaks around mask that may be bothering her and make sure she's in a comfortable position Sometimes she just wants to be turned over and repositioned and she will drift off to sleep.
- Before removing the mask, suction mouth with wand. After a long nap, secretions build up and may choke her, so doing this prevents that. If you see that the wand is not enough, prepare Deedee for deep suctioning placing her special pillow under her head/neck so she is ready for deep suction when you take off the mask.
- To remove the mask, place Deedee on her back, completely remove side straps and then lift mask off head, and take care to not scratch her with the velcro straps.
- Always remove the mask FIRST and THEN turn off the bipap. Wipe her face off and suction if needed. REMEMBER TO REMOVE THE MASK FIRST AND THEN TURN THE MACHINE OFF.
- Because she has been laying there for a period of time and can't readjust herself, she's sometime sore when she first wakes up.
- Feel free to carefully do some warming-up slow range-of-motion. She will be looking forward to some Baby Einstein or some Sing Alongs. She likes this in the mornings, this makes her start the day in a good mood.

Normal stats for Deedee are:

- Awake, with the BiPap: 98-100 /120-150HR
- Awake, without the Bipap:96-100 /130-160HR
- Sleep with the BiPap: 98-100 /90-125HR
- Sleep without the Bipap: ALWAYS HAVE BIPAP ON WHILE SLEEPING OR NAPPING TO PREVENT APNEA



WHAT TO DO WHEN DESATS OCCURS

Desats occur normally when baby chokes on her own secretions. If you don't take care of this right away, a respiratory arrest may occur. You must react FAST to any sign of baby choking. Her life depends on it.

Deedee's coughing and desating becomes more serious and more taxing on her each second that passes. You must be QUICK and do the following EFFICIENTLY.

Trust you knowledge, remain calm, and do what must be done ALWAYS reacting to how Deedee looks. Don't trust machines, trust Deedee's facial expression and skin/lips/gum color changes.

The rules are: Look at Deedee, then React... FAST!

ALWAYS notify mom or dad when Deedee's saturation is staying below 94%. It is important not to underestimate how fast a situation can shift into an emergency.

When O2 Sats below 94%:

- Suction (oral & nasal). Place special pillow inder neck, deep suction with a #10 red rubber catheter using saline bullets
- Give a few BIG Breaths with the Ambu Bag and Room Air
- If not better, Cough Assist
- Continue Cough Assist and Suction until O2 Saturation is above 94%

When O2 Sats in upper-mid 80's:

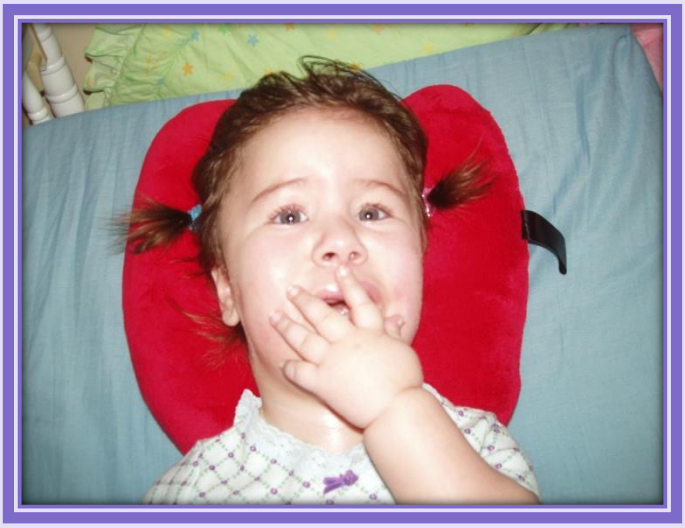
- Deep suction
- Give a few BIG Breaths with the Ambu Bag and Room Air
- If not better, use Cough Assist. Do rotating Cough Assist sets, 1 set on her right side and then 1 sets on her left side. Continue this cycle until she is above 94%
- Nasal and deep suction after each set. Use saline bullets to clear and lubricate.
- Do a full respiratory treatment and/or manual CPT/vibrations

When O2 Sats go below 80%:

- Deep suction
- Give a few BIG Breaths with the Ambu Bag and Room Air
- Move to where the Cough Assist is and start Coughing
- Include Oxygen (1-2 Liters) with Ambu. O2 is in the portable O2 tank to left of Deedee's bed. Ambu's cable is ready to plug it into the O2 tank.
- Nasal and deep suction after each set. Use saline bullets to clear and lubricate.
- Do a full respiratory treatment and/or manual CPT/vibrations

When O2 drops below 60% and she does not come right back up,

- Continue Ambu including O2 (8-15 Liters)
- Intercalate a Cough Assist set with Ambu and suction
- Nasal and deep suction after each set. Use saline bullets to clear and lubricate
- If needed, do manual CPT on lungs to get things moving immediately.
- Call 911, stay calm and focus on continuing a steady Ambu with O2 until help arrives



Note:

- Dr.Bach's medical order is to give Oxygen when saturation reaches 0, and/or there is a respiratory arrest. We wont submit Deedee to more stress than she already has during an episode, so the Ambu should be the item giving Oxygen at 2L if she is below 80% for more than a minute.

- Keep in mind that if you are massaging her foot or leaning on her foot, or moving her, her foot probe might not read accurately and so the alarm will set off. Always observe Deedee before making anything else. Her facial expression and sounds will tell you if she is in distress.

- Deedee is a belly breather, she CANNOT be seated. Please keep this in mind, it is best for her to lay down flat, especially if in distress.

- If she is coughing, or her face shows distress, immediately start with oral suction, follow with ambu, and then if needed use Cough Assist. Proceed until she is ok.

- If Deedee needs to go to a hospital, remember she must go to **University Hospital Pediatric Intensive Care Unit** as they know how to treat kids following the Non-Invasive protocol for SMA, and they know her case. After calling 911, call PICU@UMDNJ (973) 972-3784 The attending doctor should be either Dr. Nevado, Dr. Sinquaii, or Dr. Vaughan. They will be in contact with Dr. Bach.

THE DAILY BATH

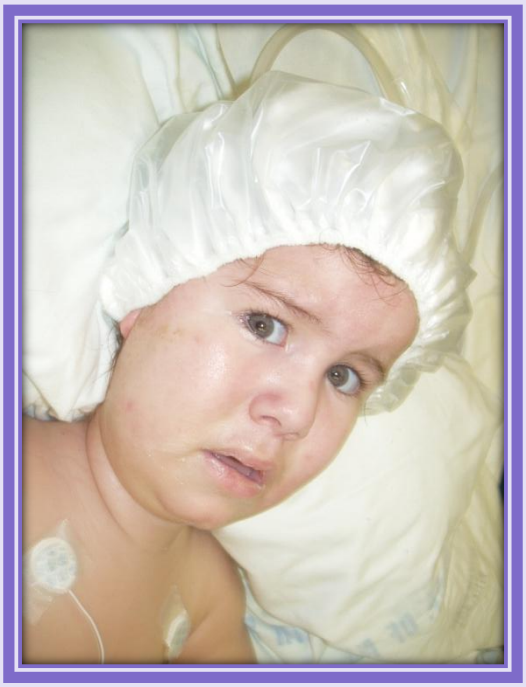
Always keep the pulse-ox on. We change it usually between 1-2 weeks (when it stops being sticky). During tub bath, we place the pulse-ox in one of Deedee's ears so it doesn't get wet.

There are two options to bath baby: Tub bath (using her bathing chair), and bed bath.

Tub bath:

- Fill up the tub until the warm water reaches the tub's limit. Place the yellow chair inside, and pur bubble bath liquid soap if using it.
- Take baby to the bathroom placing her on her wagon and then dragging it to the bathroom.
- Place the pulse ox in her ear and snap the cable and the Oximeter securely in the bath's curtain rod so it doesn't get wet. Strip baby off her clothing, and gently take her from the wagon placing her in the yellow bathing chair.
- Use the inflatable pillow for support of her head and behind her neck. Remember that baby should never be left unsupervised on the tub.
- Do range of motion exercises while baby is in the water. She moves by herself and actually exercises even more.
- Read bath book and play with toys
- Do mouth/speech therapy
- Bath baby. Use cloth with baby soap to bath the whole body naming the different body parts.
- Wash her hair (only 2 times per week). Try not to wet the pulse-ox sensor.
- Shower soap away when finished.
- Throw a towel on baby to dry her off. Then place the towel on the wagon and place baby on it. Cover baby with the rest of the towel and take her to her room. Place her in her crib.

Bed bath:



- ♥ Get ready. Prepare 2 disposable changing pads, 2 bowls (green and clear) with warm water, cloth and baby bath soap (4 drips in the green bowl).
- ♥ Place the 2 pads under baby in her crib, take off diaper cleaning off any mess, wet cloth and pass it through baby's body.
- ♥ Use the soap wash to bath baby while naming body parts and playing with her. Make sure you get the hair, back of the neck, back of ears because of the drooling.
- ♥ Rinse with water on clear bowl.
- ♥ Towel dry.

After the bath, baby gets a "spa" treatment:

- ♥ Full body massage with Cocoa & Shea Butter or Lavander Baby Lotion
- ♥ Cream up buttocks with Vaseline, apply diaper rash cream in front
- ♥ Clean up G-tube aea with saline water, the place a split gauze (if area looks red, put triple-antibiotic on it and let air dry with no gauze)
- ♥ Dress-up baby. You may let her pick up from 2 selections.
- ♥ Put Aquaphor on baby's cheeks and lips. Put also in arms and legs creases
- ♥ Bring baby to the living room, comb her hair, and start her afternoon routine

DIAPER CHANGES

Please check Deedee's diapers every two hours to see if she is wet/dirty. She typically has 1 – 2 BM diapers per day.

- ♥ We put vaseline over her butt and body creases.
- ♥ We use diaper rash cream only on her vulva.
- ♥ Do not close the diaper too tight, remember she is a "belly breather".

If she goes for longer than 24 hours without a bowel movement (BM), let us know as this can cause breathing problems (usually prune juice helps, but we keep baby suppositories in storage and we give Miralax on a feeding at a daily basis).

Extra clothes/sleepers, diapers and equipment are in Deedee's closet and under her bed. Please change her if she soaks through her diaper or if her drooling is quite excessive. We prevent her getting soaked from drool using a small diaper (size1) placed on her face while she is positioned sideways. We call it "baba collector" and it works better than cloths and gauzes.

FEEDING SPECIFICS

SMA kids should NEVER go more than 4 hours without food since many children with SMA have a fatty acid metabolism disorder and can have complications if they go too long without nutrition. They lack good fat storage to help with their energy.

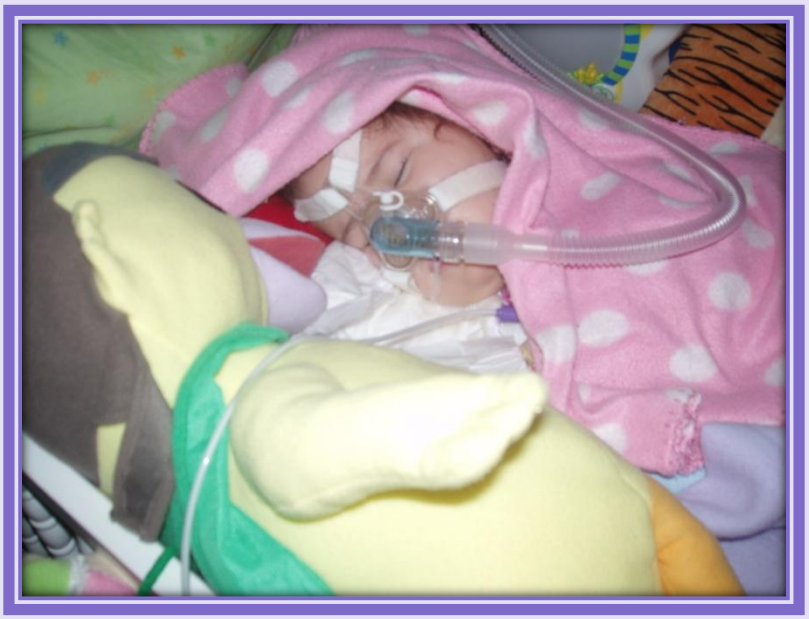
Deirdre is currently following the Amino-Acid Diet (AA diet) using ELECARE. She needs nothing else as this is an all-inclusive formula. This regimen began on September2, 2008. The goals of the AA diet include: weight loss so her lungs and movements improve, better use of nutrients and energy, discourage storage of excess water in her body. Prior to Elecare, Deedee used Similac with Iron at the same rate without any problems. Her growth and weight were fine for a normal child her age, but she is not "normal" and her weight must be watched for improvement of her quality of life.

- All foods and meds go in Deedee's g-tube. Flush after every use
- Do NOT heat or microwave Deedee's food. She uses the water bottle that is on the table, next to the ELECARE. That is her designated water bottle.
- Do not use tap water, always use the water from the bottle.
- Feedings should be 1 cup of EleCare (8oz) every 4 hours, a total of 6 feedings per day. (Revised on January, 2009, by nutritionist Geraldine Pierre). Stir the formula mixed with water, letting it set for 5 minutes. The formula is very thick, so this is necessary so it completely dilutes in the water (preventing a g-tube line plug)
- The feeding is 200ml of water and 3 scoops of ELECARE powder
- She also gets 6oz of Miralax powder in one of her daily feeds to keep bowel movements constant

Feedings are distributed this way:

8:00 am	200ml ELECARE at 135 hour rate
12:00 pm	200ml ELECARE at 135 hour rate
4:00 pm	200ml ELECARE at 135 hour rate
8:00 pm	200ml ELECARE at 135 hour rate
12:00 pm	400ml ELECARE at 70 hour rate

SLEEP TIME



Deedee usually starts fussing/crying around 9:00pm. That should let you know that she is tired and wants to go back to her room. After her night treatment (usually ends by 11pm) her mask is placed back on and she usually goes to sleep fast. We do some things to make her sleep comfortable:

- ♥ Prepare her surroundings for sleep. Place her on her side facing either the star or the aquarium (both light up, which she likes a lot).
- ♥ Place her friends around her, making sure the Glo-Bear and MrLyon are at eye range
- ♥ Sometimes place the paper fan in front of her mask so air makes it spin. Looking and hearing it soothers her and makes her go to sleep
- ♥ Cover baby with fleece if it is cold, if not use cloth. She gets hot really fast and doesn't need too much covering if the heater is fine.
- ♥ Baby sleeps listening to music. Turn on her CD player. Set it to continuous play.
- ♥ Turn on lights only after turning on her glowing toys as she gets scared of total darkness. They give her time for her eyes to adapt and also for her to go to sleep.
- ♥ If she is crying, lullabies calm her. If that doesn't work, check her diaper and her position.

WHAT DEEDEE LIKES TO DO

Please check the calendar or ask mom/dad what therapy/appointments we might have each day in home or center to be able to plan the day's activities. While Deedee has a disability, it's important to remember that she's not a vegetable. She is very intelligent, very curious, and she enjoys doing things that other kids her age do.

IMPORTANT! Reposition Deedee every 2 hours (on back, side laying, incline laying placing wedge underneath, postural drainage, stroller, bed, couch, floor mat).

Ideas on what Deedee likes to do:

- **TV programs/DVDS:** She REALLY enjoys musicals, especially the princesses sing-alongs. Please try to vary the programs she watches and sometimes talk to her about what she is watching. We prefer that she watches Baby Einstein so she actually learns something. Repeat play, just try to show her at least 3 different BE dvd's per day. She has some movies (Little Mermaid, Beauty & the Beast, and Fantasia/Fantasia2000 are her favorites) and she also enjoys most programs like Sesame Street, She-Ra, WonderPets, and Backyardigans.
- **Move:** She likes arms/legs exercises, so take advantage of that and do some stretching and bending. Range-of-motion exercises: Move her arms/legs about in slow movements. If you push in her legs (supporting the sides), tell her to "push them out", and slowly push them out as you hold them up for her. If you raise her arms up, guide her so she pulls them down. The more interaction we do with her, the more she will move for us. Eventually she mimics the moves, you need to be patient.
- **Toy swing:** Place the toy swing over her while she is on her mat. Attach the restrainers to her arms so she has a bit of freedom of movement and place different toys for her to reach. She can play music and other games using her sling restraints. Her favorites right now are the Mrs. Butterfly, the triangle, musical fish.
- **Read Books:** Just like other kids her age, she likes to be read to. She's very smart so let's keep educating her as we would a child without a disability.
- **Mobile:** We change her mobile's puppets height bringing them near her grasp so she can reach them. This encourages hand movement. Their brushing her face also helps with her nerve reactions. You will find that the crib has restraints so you can actually slip her arms on them. This will allow her to reach for the mobile puppets as they move, exercising her arms/hands and practicing grabbing
- **Flash cards for eye gaze communication:** We are working on picture cards with images of her favorite toys. This allows her to select what she wants via her gaze. Also this emphasizes vocabulary growth. Let's help

her continue to learn by using these and using teaching techniques so she discovers colors, textures and shapes!

- **Moving around the house in her wagon.** Deedee HATES being alone, and while one is in the kitchen or the home office it was difficult to have her around. But, some friends ordered a wagon so she could be moved within the house. The wagon allows for safely moving Deedee from her room to any place within the apartment. It has a sheep skin and some pillows underneath, so her comfort is ensured. This way she gets different points of view and also gets to be social and do something different besides being on her crib. She likes to play while in the wagon, especially to be read while on it.
- **Switch Toys.** Show her on/off while pushing buttons
- Practice Colors and textures (books, toys, color cards)
- Practice Farm Animals (books, toys, animal cards, songs…)
- Practice choices with eye gaze: Movies, toys, books
- **Music and Singing.** Think Sound of Music. Deedee absolutely loves music, and singing while moving puppets is a special treat for her. That's why she is so into the sing-alongs. Also, she enjoys lively music, (pop, alternative, even metal) as well as classical music
- Oral stimulation with tools/toys/gloves
- **Touch and hold things** – Bracelets, small stuffed animals, balls, and other toys are fun for her to look at and hold. Musical instruments, like her cymbals, are great to make her "clash" things up
- Play peek-a-boo or help her to touch things by guiding her hand toward things to see if she will simulate your behavior.
- **Moving fingers and waving** –We are encouraging her to do this and move her arms/legs as much as possible. She loves to be propped up under her mobile so she touches and feels the finger puppets in her face
- **Other ideas** – Feel free to bring your own ideas to help Deedee succeed. We will continue to come up with ideas to help her enjoy her days to the fullest.

In progress:

- **Position bed.** This will be great for trandenburg positioning during respiratory therapy. It was processed and we expect it by April.
- **Stand.** We hope that we can get a stander for Deedee. We are working on getting her x-ray and then ordering it. This may take a bit longer than the bed.
- **Aqua-therapy.** We are trying to get the right information to do that.
- **Going out and about.** Deedee loved travelling when she was a newborn, and now because of her condition she is stranded… and so are we as caregivers. We are trying to get a van with a lift so we can go anywhere with Deedee. Until then, going out for her means making a critical care ambulance appointment, and visiting doctors. We hope to get transportation soon so we can go to places with the right environment.

DAILY EXERCISES: RANGE OF MOTION

We count on caregivers to help complete the goal of doing Deedee's Range of Motion three times a day. First shift should do it while Deedee is in the bed, during the afternoon, mom or dad will do while she takes a bath. At night, the second shift should do it while Deedee is watching TV.

ARMS

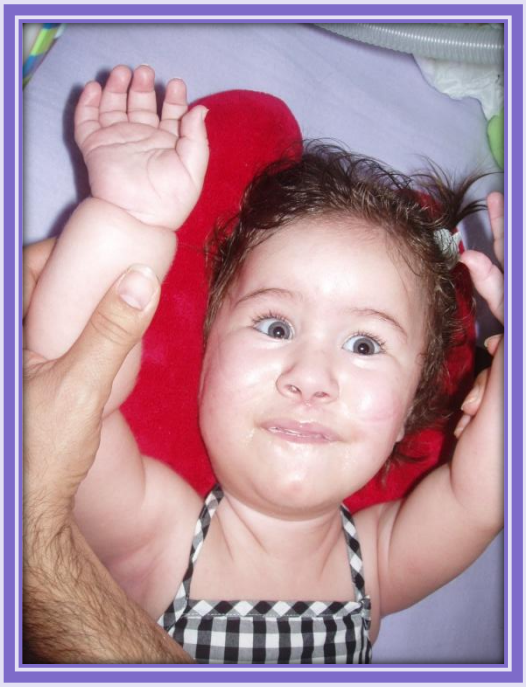
- Straight up (20 repetitions, elevator and balloon song)
- Side to side (20 repetitions)
- Up with 8 swing (20 repetitions)
- Biceps (20 repetitions)
- Wrists (20 repetitions each way)
- Fingers (stretch each)

LEGS

- Straight up (elevator & balloon song)
- Bending(bicycle) (20 repetitions)
- Thigh-master (20 repetitions)
- Ankles (20 repetitions each way)
- Toes (stretch each)

TORSO

- Twisters (side to side)



Place on the foot splints at the end of the Range of Motion routine.

JOINT COMPRESSION and RANGE OF MOTION EXERCISES LIST

Joint Compressions (3 sets of 10 on each side):

- Knee to Foot – Put one hand on the knee and one on the foot and press together.
- Hip to Knee – Put one hand on the hip and one on the knee and press together.
- Hip to Hip – Put hands on both hips and press together.
- Wrist to Elbow – Put one hand on the wrist and one on the elbow and press together.
- Elbow to Shoulder – Put one hand on the elbow and one on the shoulder and press together.

Range of Motion (3 sets of 10 on each side):

- **Foot Flexion** – Flex foot up and out to full range.
- **Ankle Flexion** – Flex foot from side to side to work ankle.
- **Kick Foot** – Lying on back, hold the leg in a 45-degree angle and bring the foot up as far as it goes and back down again. *When complete, I have Jenna do some of these on her own by asking her to kick her foot up and then bring it down.
- **Butt Lifts** – While laying on back and knees bent, place both hands under each butt cheek and lift up and then down again. *These are the same butt lifts anyone would do to work on the glutes and stretch the lower back.
- **Bicycle** – Move legs back and forth as riding a bicycle.
- Finger Extensions – Open hands all the way to stretch out fingers. Jenna has issues with opening her fingers all the way, especially the middle fingers as they lag behind. These have definitely helped.
- **Wrist Extensions**- Ben wrist up and down to full range.
- **Arm Extensions** – Bend the arm at the elbow so the arm is in a flexed position and then open it to full range.
- **Arm Punches** – Support back of the shoulder and punch the arm straight out and then back again.
- **Arm Raises** – Support the back of the shoulder and raise the arm as far as it will go over the head. I usually do this and clap her hands together or say yeah, so big, etc just to keep her attention.
- **Chicken Wing** – Support the back of the shoulder and bend the arm and lift her elbow as she is mimicking a chicken.

Head Control Exercises (DO ONLY WHEN MOM OR DAD IS SUPERVISING, DEEDEE HAS ABSOLUTELY NO CONTROL OVER HER HEAD FOR NOW, WE WANT TO HELP HER EXERCISE HER NECK BUT WITH PLENTY OF CARE. REMEMBER SHE IS VERY FRAIL!):

- **Head Raises** – While lying on back, place your arms under each shoulder blade and slowly lifer her head. Of course her head lags behind, but continue to slowly raise it to an upright and she will try to bring her head forward. These are very difficult for her to do, but she does try.
- **Sitting** – While sitting on your lap in an upright position, lightly support the back of the head and try to give less and less support.

DAILY THERAPIES

We must maintain and enhance Deedee's skills, so she can achieve as much as possible. It is necessary that on each shift, the caregiver also has a compromise towards Deedee's development. We need to encourage physical therapy, occupational therapy, speech therapy and communication therapy. All of these must be applied by everyone at different times during the day so they are actually effective.

Physical Therapy:

- ♥ Range of Motion in the morning, during bath, and at night

Occupational Therapy:

- ♥ Practice grab, let go, open, shut, push. Touch and hold things. Expose baby to different textures and surfaces. Help her touch them, reach them.

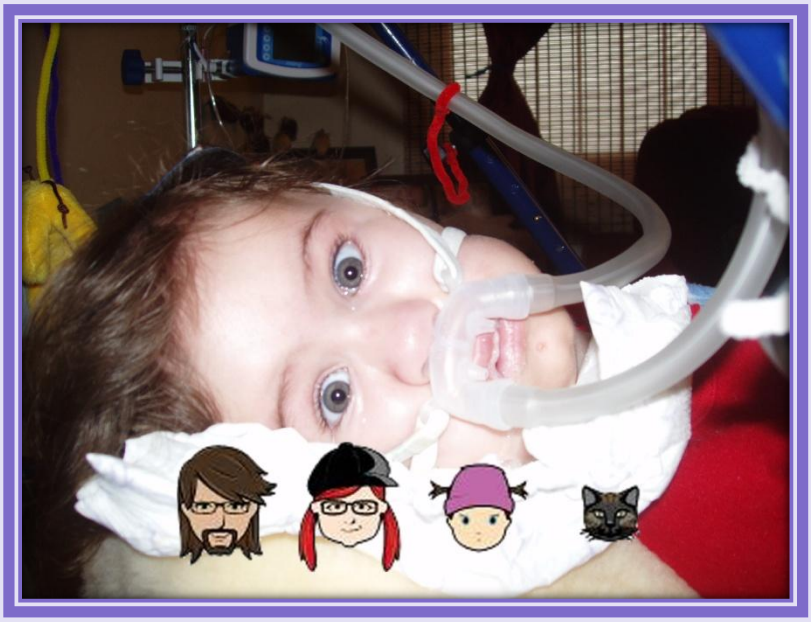
Speech Therapy:

- ♥ Vibrations around mouth and inside mouth using the vibrating toothbrush
- ♥ Stimulation of mouth by using toothettes, finger nuk, nuk. All can be covered with different flavors to add interest and diversity to baby's experience
- ♥ Stimulation of tongue using nuk and finger nuk
- ♥ Encourage repetition of vowels, repeat what she says and do sing alongs

Communication therapy:

- ♥ Choices. Give baby 2-3 choices so she selects what she wants.
- ♥ Clicker/switch. Emphasis of cause-effect through adapted toys.
- ♥ Emphasis of more and done. Flash cards with emphasis on items names, colors and shapes

DOCTORS APPOINTMENTS



Deedee needs to keep up with many doctors appointments to make sure she is fine. Each time that there is an appointment, the nurse on shift must be available 3 hours prior the appointment. This is so baby can get her full respiratory treatment session (which takes up to an hour) and to prepare her, and her things. Transportation usually arrives 45 minutes before the appointment's time.

We try to make sure that appointments are after 10:30am so there is no big changes to baby's morning schedule. If an appointment is earlier, we ask the assigned nurse to come at the right time to be of real help. Teamwork and communication are keys to avoid problems around this situation.

Please let us know your availability schedules so we can arrange services properly. Deedee mainly travels to Children's Specialized in Mountainview, NJ and to Doctor's Office in UMDNJ (90 Bergen St., Newark NJ 07103)

DEIRDRE's SUPPORT PHONEBOOK

DOCTORS

DOCTORS @ UMDNJ/DOCTORS OFFICE

Dr. Deborah Johnson
Pediatrician 973-972-0543
Dr. John Bach Jr
Phisician, SMA Specialist 973-972-2850
bachjr@umdnj.edu
Assistant bradykl@umdnj.edu
RT Lou Saporito
Millenium in UMD 973-714-3662
Dr. Brian Weaver
RRT-NPS 973-972-0700
973-972-6583
weaverbx@umdnj.edu
Dr. Geraldine Pierre
Nutritionist 973-972-5586
pierrege@umdnj.edu
Dr. Monteiro
GastroInterologist 973-972-5779
Dr. Aguila
Pulmonologist 973-972-5778

DOCTORS @ CHILDREN'S SPECIALIZED

HOTLINE 888-CHILDREN
888-244-5373
Appointments- Melissa x5715
Dr.MarinaVelickovic, Pediatrician x_____
Dr. MichaelArmento, Physician x_____
Technology Center, PT x_____
Dr.Nepolitan,
Ophtalmologist(Springfield) 973-376-5676

SMA DOCTORS/SPECIALISTS

Dr. Mary Schroth
Pulmonologist-Wisconsin 608-219-8914
Dr. Swoboda
Neurologist-Utah 801-585-1676
SMA NY Center, Columbia Univ.
Columbia University, NY

HOMECARE

NURSING COMPANIES AND NURSES

MAXIM 973-763-0820
Dominick, Vincent, Matt
Service
Pat
Nurse Department Supervisor
Christine
Nurses Supervisor
Maxim Nurses
Glenna Reyes 862-234-2767
Kathia Laborde 973-230-1463
Marie Nicholas 973-412-6326
Yverose Sanders 908-494-3731

MIDPOINT
Irma Camaligan 973-672-3833

MILLENIUM RESPIRATORY SERVICES AGENCY

HOTLINE 800-oxygen
800-269-9436
Deirdre gets respiratory equipment and daily care maintenance items from them.

Peter Cruz
RT at home 973-271-7955
Lou Saporito
RT in UMDNJ 973-714-3662
973-463-1883

TRANSPORTATION

LIFESTAR ambulance 973-812-7271
Always ask for Critical Care Ambulance.
LIFESTAR for med appointments
Lakeisha 973-972-8890 office
973-680-0966 pager

MEDI-TAXI 973-399-3796
Always ask for Critical Care Ambulance.

GOVERNMENT

EARLY INTERVENTION

Deborah Masey
Coordinator 973-395-8475

THERAPISTS
Patricia Van Orden
Developmental Int. 973-670-5731
schachph@umdnj.edu
Ellen Berman
Physical Therapy 973-715-2697
ellenbc@aol.com
Evelyn D'onofrio
Occupational Therapy 973-214-7512
Tracy Agate
Communication Specialist 973-763-6852
tagate@cpnj.org
Indira/
Speech Therapy ???
Indi001@aol.com
Susan Costello
Family Psychology 973-941-9046

DDD (DEPT. HUMAN SERVICES, DIVISION DEVELOPMENTAL DISABILITIES)

Kattia Caseres
Case Manager 973-648-3833
Angela Silva Jones
Supervisor 973-648-3797

MEDICAID

RSN Donna Kochka
Regional Nurse 973-648-8011
973-693-5065 fx
Madonna.f.kochka@dhs.state.nj.us21

EMERGENCY

EMERGENCY 911
UMDNJ-PICU
DrSinquee, DrNevado, 973-972-3784
Deirdre must ALWAYS be taken to UMDNJ PICU as they know SMA

SMA REFERENCES

We encourage any caregiver for kids with neuromuscular disease to read:
Management of Patients with Neuromuscular Disease by John R. Bach, MD

You should also know about the **Consensus on Standards of Care for Patients with Neuromuscular Disease** that was published in December's MDA Magazine QUEST (printed in the Appendix of this handbook for ease of reference).

On the web:

- The Center for Noninvasive Mechanical Ventilation Alternatives and Pulmonary Rehabilitation at UMDNJ (Dr. Bach)
  Http://www.theuniversityhospital.com/ventilation/index.shtml
- University of Utah, Department of Neurology, Pediatric Motor Disorders Research Program: SMA (Dr. Swoboda)
  http://medicine.utah.edu/neurology/research/swoboda/sma/index.htm
- Families of SMA
  http://www.curesma.com/
- MDA: Clinical Care Guidelines (Consensus of Care for SMA Patients)
  http://www.mda.org/publications/quest/extra/qe14-6_sma_guidelines.html
- Clinical Trials for SMA
  http://clinicaltrials.gov/search/term=%22Spinal%20Muscular%20Atrophy%22
- NINDS (National Institute of Neurological Disorders and Stroke)Spinal Muscular Atrophy Information Page
  http://www.ninds.nih.gov/disorders/sma/sma.htm
- NORD (National Organization of Rare Disorders)
  http://www.rarediseases.org/search/rdbdetail_abstract.html?disname=Werdnig%20Hoffman%20Disease
- SMA Medical Supply
  http://www.smasupply.com/welcome.html
- Medline Plus:SMA
  http://www.nlm.nih.gov/medlineplus/spinalmuscularatrophy.html
- Stem Cell Action
  http://www.genpol.org/
- Abnormal Breath Sounds (with sounds you can listen to so you learn to identify them)
  http://www.rnceus.com/resp/respabn.html

SMA Friends in the web:

- Shira http://www.asonginthisworld.com/index.php
- Emily http://blog.hopeforemily.com
- Ethan http://www.our-sma-angels.com/ethan
- Mary http://www.miracleformary.blogspot.com
- Viviana http://www.viviannamadera.com
- MJ http://www.our-sma-angels.com/Margaret

APENDIX 1: SMA INFORMATION

SPINAL MUSCULAR ATROPHY: EXPLAINED

Kathryn Swoboda, MD
University of Utah School of Medicine
Department of Neurology
30 North 1900 East, Room 3R413
Salt Lake City, UT 84132

SMA CHOICES

Mary Schroth, MD
University of Wisconsin American Family Children's Hospital
Respiratory Care of SMA and Choices
Revised 030908

SMA & NIV (Non-Invasive Ventilation)

John Bach Jr., MD
Professor of Neurosciences
University of Medicine & Dentistry of New Jersey (UMDNJ)
Practice Phone: Voice: 973-972-2802
Fax: 973-972-2825
Email: bachjr@umdnj.edu

MORE SMA INFORMATION

Various sources

NUTRITION: A NEW UNDERSTANDING

Richard I. Kelley, M.D., Ph.D.
Kennedy Krieger Institute and the
Department of Pediatrics, John Hopkins University School of Medicine
Baltimore, MD 21205

WHAT IS THE AMINOACID DIET?

Mary Botzo
FSMA

SPINAL MUSCULAR ATROPHY EXPLAINED

WHAT IS SMA?

Spinal muscular atrophy is a degenerative problem that affects the motor nerves, resulting in muscle wasting and weakness. Spinal muscular atrophy occurs in approximately one in 6,000 -10,000 live births.

WHAT CAUSES SMA?

SMA is an autosomal recessive disease. This means that two abnormal copies of the gene, one inherited from each parent, are necessary to have the condition. Boys and girls are affected with equal frequency. A child who inherits only one abnormal gene copy is a carrier, and is not a risk to develop symptoms.

A gene called survival motor neuron (or SMN) is found to have an abnormal area (called a deletion) in over 95 percent of cases of SMA. Symptomatic individuals of all ages can be tested through DNA studies typically done from a blood sample.

When both parents are carriers, there is a one in four, or 25 percent, chance, with each pregnancy, to have a child with SMA. Carrier testing of parents can help determine the recurrence risk in a specific family.

WHAT ARE THE SYMPTOMS OF SMA?

Spinal muscular atrophy is sometimes difficult to diagnose, as symptoms can resemble other conditions or medical problems. Each child may experience symptoms differently. There are several types of spinal muscular atrophy based on symptoms and age of onset.

- **Type I (Werdnig-Hoffman)**
  This is the most severe type of SMA, and unfortunately, the most common. Symptoms may be present at birth or develop within the first few weeks or months after birth. Infants have difficulty holding up their head, sucking, feeding, swallowing, and often move very little. The legs are more severely affected than the arms. The muscles of the chest which help to expand the lungs are affected, and the chest may appear small or "bell-shaped". They have a weak cough, and are prone to respiratory infections. The tongue may demonstrate "worm-like" movements, and they may demonstrate a tendency to choke while feeding. Complications from breathing problems often lead to death or dependence on some form of respiratory support by 2 to 3 years of age.

- **Type II (intermediate form)**
  This form of SMA most commonly becomes evident in children between 6 months to 2 years of age. They may show delays in acquiring motor skills such as rolling, sitting or crawling. They are unable to walk independently without support. They typically have generalized muscle weakness and may require braces, walkers, or a wheelchair for assistance. Life-expectancy varies greatly in this group of children, since they demonstrate a very wide range in degree of weakness. However, complications commonly include weakness of chest muscles involved in breathing, resulting in a weak cough and tendency for pneumonia. Scoliosis develops in virtually all children at some point, and they are prone to bone fractures. Children who are unable to bear weight often develop hip dislocation. Contractures of the muscles and joints can limit function over time. Children in this group may also demonstrate difficulties in swallowing and chewing, and require close monitoring of nutrition. Lifespan in this group depends on the severity of respiratory muscle weakness, but many children survive well into adulthood.

- **Type III (Kugelberg-Welander)**
  This form of SMA most commonly becomes evident in children between 2 and 17 years of age. These children may show delays in motor development, difficulty walking, trouble getting up from the floor, mild muscle weakness, and frequent falls. Fatigue can be a significant problem, which limits the ability to walk long distances. A tremor involving the hands is common. Scoliosis is frequent in later childhood. Respiratory muscle involvement is much less often a problem, and difficulty swallowing is rare.

- **Type IV**
  This form of SMA includes those individuals who don't develop symptoms of weakness until they reach adulthood. Usually, this results in muscle weakness predominantly affecting the legs, and manifests as a walking disability. The symptoms of spinal muscular atrophy may resemble other problems or medical conditions, and can be confused with other muscle or nerve conditions, including muscular dystrophy, myopathy, other spinal muscular atrophy variants or even forms of amyotrophic lateral sclerosis (ALS). Another closely related condition, known as spinobulbar muscular atrophy (Kennedy's disease), can also present in late childhood or adulthood.

SMA DIAGNOSIS

The diagnosis of spinal muscular atrophy may be suspected if you or your child demonstrate specific symptoms or demonstrate signs on examination that are consistent with the pattern of weakness seen in this disorder. During the physical examination, your child's physician will obtain a complete medical history, and he/she may also ask if there is a family history of any medical problems.

Diagnostic tests which can help to confirm the diagnosis of spinal muscular atrophy include:

- Blood tests:
  Genetic testing can confirm a suspected diagnosis in most cases. Sometimes the initial genetic test is negative, and additional genetic testing or other testing may be needed to confirm a diagnosis.

- Electromyogram (EMG):
  A test that measures the electrical activity of a muscle or a group of muscles. An EMG can detect abnormal electrical muscle activity due to diseases and neuromuscular conditions.

- Muscle biopsy:
  A small sample of the muscle is removed and examined to determine and confirm a diagnosis or condition.

- MRI or imaging studies:
  Imaging studies of the brain or spine to help rule out other conditions

MANAGEMENT & TREATMENT OF SMA

While there are as yet no specific pharmaceutical therapies which have been confirmed which can either extend lifespan or increase strength in SMA subjects, the identification of compounds that can increase SMN protein in cells from SMA subjects and in genetic animal models of SMA is promising. Moreover, proactive management strategies to optimize lung function, physical mobility and nutrition can help preserve motor function, improve quality of life and extend survival, particularly in more severely affected SMA infants and young children.

Following diagnosis in children with milder forms of SMA, including the infant with an as yet uncertain prognosis, it is important to work closely with parents in order to anticipate problems and pursue management aggressively to optimize outcomes. Because of the tremendous variability in severity of muscle weakness, an individualized approach is often necessary. Far too often, respiratory, nutritional and even physical rehabilitation interventions are reactive rather than proactive.

Treatment for the intermediate or milder forms of SMA should be focused on preserving mobility and minimizing respiratory complications, particularly restrictive pulmonary disease or respiratory compromise due to progressive scoliosis. Specific interventions can be helpful in optimizing the individual's health and helping to maintain motor function.

RESPIRATORY MANAGEMENT OF SMA

Since respiratory management can impact lifespan considerably, physicians should support families in implementing a proactive approach. While pulmonary management is often demanding, it is also the therapeutic modality that will most likely enhance quality of life and prolong lifespan.

To maintain lung capacity, breathing exercises and supplementary aids may be helpful. Incentive spirometry and "breath-stacking" can be implemented at an early age but require discipline to perform on a daily basis. Aerosol therapy with nebulizers may be helpful in some settings and can be initiated at the onset of respiratory symptoms.

Benefit from the routine use of mucolytics, bronchodilaters or steroid treatments is unclear and should be dictated by individual circumstances. Cough assist devices such as the inexsufflator (cough machine) are essential when ineffective cough inhibits adequate removal of bronchial secretions in the lower airways. Vest therapy or manual percussion techniques to help mobilize secretions, when used in conjunction with the cough assist machine, can be additionally helpful in some patients. Regular use of such therapies, most importantly the cough assist machine, are invaluable in the setting of a superimposed respiratory illness, and can help prevent a simple "cold" or "bronchitis" from evolving into pneumonia or a collapsed lung resulting in respiratory crisis.

Even when children are well, daily use of such a regimen can help minimize atelectasis and chest wall contractures and deformity. In many children and young adults, nocturnal hypoventilation with and without obstructive apnea necessitates assisted ventilation.

BiPAP is recommended whenever the vital capacity falls to < 40%. However, in infants and young children who can't cooperate with formal pulmonary function testing, doctors must use other signs and symptoms to help decide when a child might benefit from BiPAP support. Recurrent nighttime awakenings are often an indication that patients may not be optimally breathing during sleep. A sleep study may help to determine whether or not there is an obstructive component (due to low muscle tone or enlarged tonsils or adenoids), or whether nocturnal hypoventilation (shallow-breathing resulting in low oxygen levels or increased carbon dioxide levels) is present. BiPAP is often recommended for use only at night, but can be invaluable to use for longer periods of time when an upper respiratory infection or other illness results in increased work of breathing and fatigue.

Flu prophylaxis is recommended annually. In younger infants and children with significant intercostal weakness (all type I and weak type II subjects), prophylaxis for respiratory syncytial virus (RSV) is also recommended.

Pulmonary medicine consultation is recommended to assist in making decisions regarding long-term respiratory management. Aggressive treatment of respiratory infection is essential. Antibiotic use is of value when symptoms arising from a

presumed viral upper respiratory infection persist longer than expected or new fever or altered secretions appear in the midst of an apparent viral illness. Since recurrent or prolonged antibiotic treatment can predispose patients to yeast infections or even enterocolitis, a balanced approach is needed.

In the severely compromised infant or child, a lower threshold for administering antibiotics may be warranted. If illness results in persistent hypoxemia below 93%, the need for hospitalization and potential intubation should be discussed, although many such patients can be managed effectively in hospital using a non-invasive respiratory protocol.

Oxygen therapy should only be used in conjunction with assisted ventilation in such patients as it can suppress respiratory drive, resulting in atelectasis and hypercarbia.

View Daily Respiratory Management Strategies video at http://medicine.utah.edu/neurology/research/swoboda/videos/SMA_Respiratory.htm

ACUTE RESPIRATORY ILLNESS CARE GUIDELINES FOR SMA CHILDREN

For the not yet intubated patient:
Perform the following a minimum of every 2-4 hours, or as frequently as needed if there are copious secretions. Alternate position of patient every cycle to either side or back, guided by x-ray appearance:

1. In-exsufflator cough machine set at inspiratory pressure of + 30 to 40 cm H20 for 1-2 seconds, expiratory pressure –30 to 40 cm H20 for 1 second and 1 to 2 second pause: 4 sets of 5 breaths, followed by

2. Upper airway suctioning (avoid deep suctioning when possible, as it can result in edema and worsen the situation)

3. If there is partial or complete collapse of a lobe, intubation may be inevitable, but could potentially be reversed with continuous BiPAP support (pressures 16-20 over 4-8) and frequent respiratory PT/cough assist treatments to remove upper airway secretions.

4. Oxygen supplementation can be used with BiPAP support, but oxygen without BiPAP support should be avoided, as it will result in decreased respiratory effort, atelectasis and CO2 retention. If work of breathing is too great, with HR persistently 10-20 % above baseline, intubation may be inevitable and is best done proactively.

5. When ill, patients are remarkably vulnerable to decompensation with respiratory treatments. Pre-treatment hyperventilation with bag

and mask, and supplemental oxygen via cough assist tubing during treatments may be helpful. Be prepared to intubate as necessary; these patients have an increased propensity to demonstrate a vagal response with stress; this may resolve fairly quickly if the stimulation evoking it is stopped, and oxygen with or without bag/mask ventilation is given immediately.

6. If intubation is necessary, it is important to remember that these patients often have jaw contractures, which can make intubation a challenge.

Nutritional Management Guidelines:
It is common practice to stop feeds when monitoring these patients during illness due to the increased risk of aspiration should they require urgent intubation. However, these patients have little to no reserve of lean body mass to mobilize energy, fatigue more quickly when catabolic, and are more likely to become acidotic when adequate nutrition is lacking.

1. If fasting is expected to exceed 6 hours, recommend institution of continuous NJ feeds using an elemental or easy to digest formula containing protein.

2. If intubation seems imminent, and aspiration risk is considered too high, institute peripheral or total parenteral nutrition (PPN or TPN) with dextrose 10-15% and 1.5 grams/kg/24 hours amino acid with vitamins/electrolytes. Intralipid infusion can be used, but calories from lipid shouldn't exceed 15-20% of total calories.

3. Avoid giving just dextrose and electrolytes without amino acids for prolonged periods of time, as it frequently results in hyperglycemia and acidosis.

For the intubated patient:
Perform the following a minimum of every 4 hours (alternate position of patient every cycle to either side or back, guided by x-ray appearance):

1. Intrapulmonary percussive ventilation followed by

2. In-exsufflator cough machine set at inspiratory pressure of + 30 cm H20 for 1 second, expiratory pressure –30 cm H20 for 1 second and 1 to 2 second pause: 4 sets of 5 breaths, followed by

3. Endotracheal tube and airway suctioning

Note: the inexsufflator machine (cough assist) can be used as often as needed followed by endotracheal suctioning to help remove excessive lower airway secretions.

Extubate when patient is:

1. Afebrile
2. Not requiring additional supplemental O2
3. CXR without atelectasis or infiltrates
4. Off respiratory depressants for at least 24 hours – this includes ativan, versed, valium and derivatives
5. Necessity of airway suctioning is close to baseline

Extubate to continuous nasal or mask ventilation (without supplemental 02 or minimal supplemental O2) such as BiPAP, settings: IPAP 12 to 20, EPAP: 3 to 6 using the spontaneous mode. Use ST (spontaneous timed) mode with backup rate if patient is unable to initiate a breath with BiPAP. Backup rates are often higher than in patients without neuromuscular disease.

Use oximetry as a guide for use of expiratory aids, postural drainage and CPT.

Following extubation perform the following at least every 4 hours:

1. Chest PT manual or IPV, followed by

2. In-exsufflator cough machine, set at inspiratory pressure of +40 for 1 second, expiratory pressure –40 for 1 second and pause for 1 to 2 seconds, 4 sets of 5 breaths, followed by

3. Postural drainage (trendelenberg) up to 15 minutes as tolerated, followed by

4. In-exsufflator cough machine, 4 sets of 5 breaths, settings as in 2.
The inexsufflator cough machine can be used as often as every 10 minutes to clear lower airway secretions. Use acutely if oxygen saturation drops to < 94%. Wean from BiPAP during the day as tolerated. Goal is to use BiPAP per nasal mask while sleeping only, or to wean to their prior baseline use of BiPAP. Wean airway clearance regimen above as airway secretions decrease working toward a QID schedule or less.

Nutritional Management during concomitant illness/catabolic states is critical! Do not allow fasting > 6 hours in SMA type 1 subjects, or > 8 to 12 hours in SMA type 2 subjects or other neuromuscular patients. SMA patients have a secondary fatty acid oxidation defect and most have reflux, delayed gastric emptying and autonomic gastrointestinal dysfunction which worsens with illness: recommend immediate institution of continuous feeds via NJ, plus/minus supplementation with PPN including Dextrose 10-15%, between 1 -2 gms/kg amino acids per 24 hour period and no more than 10-15% intralipid if indicated.

RECOMMENDATIONS FOR POST-OPERATIVE CARE OF SMA CHILDREN

For the intubated patient, perform the following q 4 hours:

1. Chest physiotherapy, either manual or intrapulmonary percussive ventilation (IPV) followed by
2. Cough Assist machine set at inspiratory pressure + 25 - 40 for 1 second, expiratory pressure – 25 - 40 for 1 second and 2 second pause, followed by endotracheal tube suctioning. Pressures depend on degree of neuromuscular weakness and prior use of cough assist treatments. Do 4 sets of 5 breaths followed by suctioning after each set. G-tube should be vented during treatments.
3. If the patient tolerates, postural drainage (trendelenberg) up to 15 minutes, followed by
4. Cough Assist machine, 4 sets of 5 breaths and suctioning after each set.

Use of the cough assist machine following surgery can help to re-expand lungs and minimize atelectasis. Extubate when the patient is:

1. afebrile
2. not requiring supplemental O2
3. lung exam clear and CXR (if indicated) without atelectasis or infiltrates
4. off respiratory depressants
5. airway suctioning is 1 time/hour or less

Extubate to continuous nasal mask ventilation and no supplemental O_2 such as BiPAP with settings: IPAP 15-20 and EPAP 3-6 using the spontaneous timed mode with a backup rate to match their respiratory rate.

Use oximetry to guide the use of expiratory aids, postural drainage, and CPT, e.g., if SpO2 < 94% use Cough Assist.

Following extubation perform the following q. 4 hours:

1. Chest physiotherapy, either manual or IPV, followed by
2. Cough Assist machine, set at inspiratory pressures +25 to 40 for 1 second, expiratory pressure –25 to 40 for 1 second and 2 second pause, 4 sets of 5 breaths, followed by
3. Postural drainage (trendelenberg) up to 15 minutes as tolerated

The Cough Assist machine can be used more frequently as needed to clear lower airway secretions and reverse atelectasis. Use if oxygen saturation drops to < 94% acutely.

Wean off BiPAP as tolerated. Goal is to use BiPAP per nasal mask while sleeping only. Wean airway clearance regimen frequency above as airway secretions decrease working toward a 4x/day schedule or less.

NUTRITION

Nutrition is critically important for maintaining muscle mass and strength and minimizing fatigue in SMA patients. Intermittent monitoring of dietary intake in consultation with a dietician experienced in management of patients with neuromuscular conditions or metabolic disorders can be extremely helpful.

Children may be deficient in carnitine due to decreased intake of meat related to jaw contractures interfering with chewing. Swallowing problems sometimes be difficult to detect, and in weaker children or in those with borderline nutritional status or frequent respiratory illness, swallowing studies should be performed on a regular basis to ensure that silent aspiration isn't contributing to respiratory problems.

Maintenance of appropriate nutrition is especially critical during illness. SMA subjects have diminished lean body mass and a secondary defect in fatty acid oxidation that limits their reserve in the setting of prolonged fasting. Thus, when concerns regarding the ability to safely administer oral feeds develop, alternative forms of nutrition should be considered. Options include temporary nasogastric or nasojejunal feeds, or peripheral or total parenteral nutrition (PPN or TPN). Check the General Nutrition Guidelines for SMA Children table on the next page.

Guidelines for Gastrostomy Tube Feeding for Infants with SMA type I

Formula Options: At this time, there is no good data on whether a specific brand or brands of regular infant formulas are better tolerated in infants with SMA type I. Even after infants get a gastrostomy tube (g-tube), they can continue to benefit from receiving breast milk, which contains valuable proteins to help bolster an immature immune system. If the infant still has the ability to safely feed orally as documented by a swallow study (but perhaps received a gastrostomy tube due to poor weight gain or a single aspiration event during illness), he or she can sometimes still be breast or bottle fed, but supplemented additionally as needed with breast milk or formula via g-tube. In this case, weekly monitoring of weight gain can help guide appropriate g-tube supplementation.

Obviously, infants who choke or sputter with feeds are at risk of aspiration, and feeding should be restricted to g-tube only.

Elemental formulas are easier to digest because they contain free amino acids rather than whole proteins; however, they don't taste good, are much more expensive, and are not clearly superior to regular formulas for those infants who tolerate them. Elemental formulas with lower fat content may be better tolerated than those with higher fat content, such as **Neocate**. Many babies have abnormal gastric emptying, and a lower fat, elemental based formula such as **Vivonex** may be more ideal for those babies demonstrating reflux or other difficulties tolerating feeds. Since babies with SMA have a lower lean body mass, lower metabolic rate and thus lower calorie requirement, the quantity of formula should be adjusted as needed to meet the baby's growth requirements, with a goal of maintaining weight

General Nutrition Guidelines for SMA Children

Calories
8-11 kcal per centimeter height (conversion is height in inches X 2.54) Ideally, caloric intake should be monitored and adjusted as needed to maintain weight for height ratio around the 10th percentile. This is because SMA subjects have significantly reduced lean mass.

Protein
Recommended range is 1 - 2 grams per kilogram body weight (conversion is weight in pounds divided by 2.2). Protein in excess of 2 grams/kg/day over a long period of time could potentially result in kidney problems and negatively affect bone density.

Fat
Try not to exceed 30% of total calories. Children less than age 2 years require a critical minimum amount of fat in the form of essential fatty acids for normal brain development. Please consult a dietician if you have specific questions regarding such guidelines.

Fiber
A good general guideline is age plus 5 (a 3 year old would need 3 + 5 = 8 grams of fiber a day)

Fluid
115 – 135 ml/kg (conversion is weight in pounds divided by 2.2). Children with fever or excessive fluid loss from sweating may require more in some cases.

Vitamins and Minerals*

Nutrients	1-3 yr old	4-8 yr old	9-13 yr old	*14-18 yr old
Vitamin D	5 mcg	5 mcg	5 mcg	5 mcg
Vitamin A	300 mcg	400 mcg	600mcg	900/700 mcg
Vitamin E	6 mg α-toco	7 mg α-toco	11 mg	15 mg
Vitamin C	15 mg	20 mg	45 mg	75/65 mg
Thiamin	0.5 mg	0.6 mg	0.9 mg	1.2/1.0 mg
Riboflavin	0.5 mg	0.6 mg	0.9 mg	1.3/1.0 mg
Niacin	6 mg	8 mg	12 mg	16/14 mg
Vitamin B6	0.5 mg	0.6 mg	1.0 mg	1.3/1.2 mg
Folate	150 mcg	200 mcg	300 mcg	400 mcg
Vitamin B12	0.9 mcg	1.2 mcg	1.8 mcg	2.4 mcg
Vitamin K	30 mcg	55 mcg	60 mcg	75 mcg
Calcium	500 mg	800 mg	1300 mg	1300 mg
Iron	7 mg	10 mg	8mg	11/15 mg

*Where two numbers are listed, the first number refers to the recommendation for males, the second is for females.
mg = milligram
mcg = microgram
IU = international unit
a-toco = alpha-tocopherol

***These are recommended daily intakes, not tolerable upper limits. It is not known whether or not these recommendations are ideal for SMA subjects , but can provide a guide to appropriate intake. Certain vitamins in excess, such as B6 (pyridoxine), can be toxic to nerves. Niacin in large quantities can cause flushing and stomach upset. Thus, it is wise to be conservative in providing vitamin supplements greatly in excess of RDA requirements.**

Last updated 11/2007. Prepared by Kathryn Swoboda, M.D. and Gail Wiebke, R.D., University of Utah to help promote minimum standard of care guidelines for nutritional management in infants and children with SMA. These recommendations are guidelines only; please consult with your physician regarding specific care issues in you or your child. For questions regarding this document, you may email Swoboda@genetics.utah.edu.

for length ratio of 10-25% (See SMA Nutrition Guidelines). **Constipation** is frequent in infants with SMA type I, and can add to abdominal bloating and discomfort and worsen reflux. Regulation of bowel movements with glycopyrrolate or a promotility agent such as metoclopramide may be indicated if they aren't having at least one or more bowel movements every day.

Feeding Strategies: Children with SMA type I may have some difficulties tolerating bolus feeding, so that smaller more frequent bolus feeds during the day may be best, every 3 hours or so, especially if they don't have a Nissan. All babies spit up (have reflux), but with SMA babies this is much more frequently a problem, particularly if their ability to protect their airway is compromised. Proactive respiratory care using a cough assist machine or BiPAP can allow some air into the tummy, which can further aggravate the situation. Be sure to vent the g-tube during treatments with the cough assist, and when on BiPAP. Keep the baby's head and body up at least at a 30 degree angle from horizontal for at least 30-60 minutes following a bolus feed. A wedge under his/her mattress or play area works well, or a seat that is angled so that the tummy isn't compressed. Don't assume he or she won't tolerate regular formula until you try it, as some babies tolerate regular formula without difficulty. If he or she fails to tolerate the regular formula with bolus feeds, a continuous feeding regimen using a pump may be helpful, or smaller bolus feeds during the day in combination with continuous nighttime feeds. Finally, if those strategies fail, we recommend substituting an elemental formula which is lower in fat, such as pediatric Vivonex.

PERI-OPERATIVE NUTRITIONAL CARE GUIDELINES FOR SMA CHILDREN

Nutritional Management during catabolic states is important to minimize fatigue and prevent respiratory failure for children with neuromuscular disease undergoing elective surgical procedures. Their diminished lean body mass limits mobilization of amino acids during prolonged fasting or illness. In the peri-operative setting, it is common to restrict feeding after midnight the day prior to the procedure. In the case of a prolonged procedure, such as scoliosis surgery, this can mean that children will have restricted oral intake for 24 -36 hours. This is subsequently followed by the slow introduction of nutrition, starting with clear liquids. Some children may experience nausea due to the medications they've received during the procedure or afterwards to treat pain related to the procedure. Under usual circumstances, children typically receive intravenous fluid with sugar during the recovery period. However, the amount of calories received is limited, and children with neuromuscular disease appear to be more likely to develop hyperglycemia (higher blood sugars) in this setting. Finally, many neuromuscular patients have abnormal gastric motility and reflux which can be exacerbated in the post-surgical setting.

Prolonged fasting or inadequate caloric intake can negatively impact recovery by enhancing fatigue and making it more difficult for them to wean off ventilatory support. Under the most ideal circumstances, pre-operative fasting should be

limited to no more than 6 hours in SMA type I subjects or weak neuromuscular infants, and to no more than 8 hours in SMA type II or type III subjects or other neuromuscular patients.

Recommended NPO times
6-8 Hours before procedure (depending on child's age): As per anesthesia guidelines – No solids, formula or non-breast milk allowed. Clear liquids and breast milk are still acceptable.

4 hours before procedure:-As per anesthesia guidelines-Clear liquids only from this point forward. Children be encouraged to drink fluid up until the 2 hour limit. Clear liquid could be a beverage that contains glucose as well as amino acids.(see below). Other acceptable but clear liquids include water, pedialyte, apple juice, Gatorade, or clear soda such as Seven-Up or Sprite.

2 hours before procedure- As per anesthesia guidelines-No more intake prior to surgery after this time point.

Clear Liquid Alternative for Neuromuscular Patients:
Resource Fruit beverage or Boost Breeze are two options (parents can go to www.resource.walgreens.com and look under before/after surgery medical nutrition tab). Other comparable clear liquid products include Nestle Carnation Instant Breakfast Juice Drink www.nestle-nutrition.com

During Operative Procedure
An intravenous line (I.V.) will be placed prior to the procedure, sometimes in the pre-operative waiting area and sometimes in the operating room. The doctor or nurse may check a blood sugar level and other laboratory studies at this time. Depending on the length of the procedure, intravenous fluids with or without sugar will be administered to maintain hydration.

Post-Operative Management
Once your child has been taken to the recovery room, peripheral parenteral nutrition (PPN) can be started through the I.V.. The intravenous solution used will include dextrose 10-15%, 1.5 gms/kg amino acids per 24 hour period and standard vitamins, minerals and electrolytes. This will provide a source of sugar and amino acids to help maintain your child's energy level, and keep blood sugar levels stable. If oral intake is restricted for more than 48 hours, the dietician may recommend adding a 10% intralipid infusion. However, since SMA patients have a secondary defect in fatty acid oxidation, total fat from all sources should not exceed 15-20% of total calories (all intralipid infusions available in the U.S. currently contain long-chain fatty acids). Peripheral parenteral nutrition (PPN) will need to be ordered in advance the morning of the procedure in order to be ready during the post-operative period. Most pharmacies require several hours notice to prepare these solutions. Total peripheral nutrition, or TPN, requires a larger IV, and allows even more sugar and fat to be administered to help boost calories. However, this is usually not necessary in an uncomplicated peri-operative setting.

Recommendations for oral re-feeding:
Once a child begins taking clear liquids, PPN or TPN can be discontinued. However, if nausea or decreased appetite limit intake, PPN can be used for several days to enhance the recovery process. The clear liquid beverages which contain hydrolyzed protein can also be used in this setting to provide an additional protein source if the child is unable or unwilling to advance to regular feeds. If the child has a g-tube, he or she can begin receiving an elemental or semi-elemental formula, like pediatric vivonex, or tolerex (double-diluted), if regular formulas are not tolerated. These formulas are sometimes better tolerated than regular formulas like pediasure, as they have lower fat levels, and help facilitate more rapid gastric emptying.
The goal should be to advance oral supplementation to estimated full caloric requirements based on body weight/length for neuromuscular patients (see document entitled: general nutritional guidelines for neuromuscular patients) no later than one to two days post-operatively, or sooner if possible.
For difficulties in weaning children from mechanical ventilation, see "POST-OPERATIVE RESPIRATORY CARE GUIDELINES FOR CHILDREN WITH SMA" in the Nutrition section.

EXERCISE AND MAINTENANCE OF MOBILITY

Physical therapy on a daily basis, performed by parents with appropriate direction from a physical therapist, can help to minimize joint contractures and maintain mobility. The use of standing devices, appropriate orthotics, bracing, and facilitated ambulation can help significantly in this regard. Weight bearing on a daily basis, instituted as early as possible in non-ambulatory children for a minimum of 2 hours per day, can help to delay onset of scoliosis, limit contractures, and improve circulation and gastrointestinal motility.

Daily exercise should be encouraged, to include the upper extremities in those children with severely impaired lower extremity function, to help maintain motor function and promote cardiovascular health. A daily exercise program could include part-time use of manually propelled mobility devices, including manual wheelchair, manual stander, or gait trainer.

The major orthopedic problems these children face include scoliosis, hip dislocation; and an increased risk of fracture due to decreased bone density and propensity for falls. The age of onset and rate of progression of such complications are directly related to the severity of muscle weakness, but early intervention can minimize the impact and severity of such problems.

Contractures can develop quite rapidly in the setting of illness, excessive time spent in a wheelchair, decreased activity, or recovery after scoliosis surgery or other orthopedic procedures. Daily range of motion and early return to supported weight-bearing can help maintain function in these children.

Helpful links: www.mobilityfordiscovery.com www.standingdani.com Panthera wheelchairs for smaller children up to 30 kg, consider the Panthera micro www.childrite.com

GUIDELINES FOR ACTIVITY FOR CHILDREN WITH SMA WHO ARE AMBULATORY, WITH OR WITHOUT BRACES OR ASSISTIVE DEVICES

Background:
SMA is associated with decreased lean body mass. In stronger SMA patients, muscle weakness most noticeably affects the hip and shoulder girdles, making it more difficult to arise from the floor without assistance, and sometimes associated with a somewhat wide-based unsteady gait. Joint contractures (decreased range of motion around a joint) are much less common in ambulatory SMA children. However, development of contractures can occur during periods of decreased activity or illness. In ambulatory children, contractures most commonly affect the hips and ankles. Associated tightness of the hamstrings also often occurs in this setting. Toe-walking is often a sign of developing ankle contractures. Ankle contractures can lead to an increased probability of tripping, since tightness of the Achilles tendon can limit how far the foot can flex upward to clear the ground. Tightness of the hips over time, in association with weak abdominal muscles, can lead to an enhanced lumbar lordosis, or "swayback" position. Muscle weakness, particularly of the quadriceps muscle group which helps to stabilize the knee, can also contribute to an increased risk for falls.

Importance of Stretching:
Development and progression of lower extremity contractures can be prevented or delayed with a combined exercise and stretching program. Stretching should target the hips, thighs, low back, hamstrings and ankles on a daily basis if possible. Building this into a child's routine is important to establish early. Learning to stretch properly under the guidance of a physical or occupational therapist is best. The frequency with which such stretching can be incorporated into the daily and weekly routine is a primary key to success. Performing such routines following exercise, after a warm bath, or just before bedtime can help maximize benefit and acceptability. Sometimes just a few days in bed can weaken the quadriceps sufficiently due to muscle atrophy to really impair ambulatory capacity and result in dangerous falls. Don't forget the daily stretching, even if your child is ill or recovering from surgery, and early ambulation is advisable to help maintain function.

Physical Activity:
Just as important as stretching, or even more so, is some form of regular exercise to help maintain muscle strength. Daily physical activity is important, and this can be supplemented with more specific focused therapy with your PT or OT to target weak muscles and work on specific functional goals. Using a wheelchair to minimize fatigue over longer distances is fine. However, encourage them to stand or walk on their own at home and at school as much as possible. Stairs can be viewed as

exercise opportunities rather than obstacles (with proper technique and monitoring!). Cycling, swimming and walking are excellent sources of aerobic exercise. However, be creative and have fun! Tai Chi or dance can provide children with a great experience that incorporates elements of stretching, balance, strengthening and activity in a setting where self-image is likely to be enhanced. Use of light weights with no more than moderate repetition frequency can help maintain strength in the upper extremities. Sit-ups or other abdominal strengthening exercises – with or without an exercise ball or peanut, and performed following proper instruction by your therapist to avoid back injury, can be helpful in maintaining good posture and minimizing back pain. The most important thing is to find a routine that works for you and your child. They will be much more likely to want to participate if it is fun.

ORTHOPEDIC ISSUES

Scoliosis almost invariably begins in the first decade of life in SMA type II and in a substantial proportion of children with SMA type III. The curves progress over time, sometimes quite rapidly during transition to increased wheelchair use or in conjunction with a growth spurt. In non-ambulatory patients, spinal bracing may improve sitting stability, as long as care is taken not to compromise abdominal movement in those with intercostal muscle weakness.

However, continuous use of such bracing should be limited if possible in order to maintain trunk strength and mobility. Periodic pulmonary function studies help to establish a profile for the individual patient, allowing design of the most appropriate care plan surrounding respiratory care in the post-surgical period. Since worsening is inevitable in most children, once the curve reaches 40 degrees a decision to intervene may be warranted. When very young patients develop scoliosis, bracing can sometimes help to defer surgery for variable periods of time, and "growth rods" or other means of accommodating growth may be indicated.

Proximal muscle weakness predisposes patients to progressive subluxation and dislocation of the hip. Subsequent hip degeneration can result in significant chronic pain. In non-ambulatory patients, it is important to prevent the hips from dislocating for reasons of comfort, good sitting balance, and maintenance of pelvic alignment.

To achieve an optimal result, operative intervention may be required in some cases. Patients who have type III SMA and are still able to walk present a difficult management problem. These patients are also prone to subluxation of the hip due to significant proximal muscle weakness. However, because surgical intervention with proximal femoral varus osteotomy may result in additional weakening of the abductor muscles, the physician should be cautious in recommending such surgical procedures in an ambulatory patient (Vertical Expandable Prosthetic Titanium Rib) (VEPTR)

Since these patients rely to a great extent on lumbar lordosis and a side-to-side waddle to walk, bracing or spinal arthrodesis may worsen their gait. It is not uncommon for SMA type III patients to become non-ambulatory following spinal surgery, particularly if a rehabilitation plan is not instituted immediately in the post-operative period. Joint contractures can progress quite rapidly in this setting without dedicated prevention. In a subset of cases in which ambulatory status is considered at risk, postponement of surgery may be the best choice. Special consideration of nutritional support in the perioperative period can help ensure a good outcome.

SPECIAL CONSIDERATIONS IN SMA INFANTS

Before and after confirmatory genetic testing is completed in an obviously severely affected infant, it is essential to work closely with parents to ensure that they understand what they may face in the months following the diagnosis, fully reviewing their options regarding supportive nutritional and respiratory interventions. This is particularly important to address early, since these infants are often diagnosed in the setting of an initial respiratory event and may already have bulbar insufficiency and respiratory insufficiency.

It is important that families be presented with a range of options and that quality of life for the entire family be preserved as much as possible. Many parents, when presented with options, choose to forgo invasive diagnostic and therapeutic procedures. However, others are anxious and willing to embrace a very proactive care plan if it means extending the life of their child.

It is vital to maintain open communication so that all caregivers are aware of choices already made as well as areas of continuing uncertainty regarding interventions to be considered.Proper positioning, daily passive range of motion, and use of alternative mattress systems or seating devices can enhance quality life. A flat car bed rather than a car seat is advisable for infants who rely primarily on abdominal breathing. If swallowing problems are mild, thickening the formula and positioning the infant properly can help to avoid aspiration of formula into the lungs.

However, families of Type I infants will almost certainly need to consider some alternative means of providing nutrition at some point. A nasogastric or nasojejunal tube (a slender tube which goes through the nose and down to the stomach or to the first part of the intestine) is often suffcient for prolonged periods of time, allowing the family time to consider the range of options.

More permanent options include gastrostomy or a combined Nissan/gastrostomy procedure. While surgery and general anesthesia clearly carry some potential risks, laparoscopic techniques are available that allow a more rapid recovery. In severely weak infants in whom general anesthesia is a concern, percutaneous gastrostomy with local anesthesia is an option. Given the SMA child's need to eat regularly,

limiting fasting prior to such procedures and providing nutritional support immediately afterward will help to enhance recovery.

Respiratory management is a challenge, presenting the greatest risk of death or serious disability in the weakest infants and children. The most aggressive approach, including tracheotomy and mechanical ventilation, does nothing to prevent disease progression. Complications such as tracheitis (infection involving the windpipe), sepsis (life-threatening infection in the blood) and ongoing respiratory complications can compromise quality of life.

Families may be pressured to make quick long-term decisions without adequate preparation when infants are intubated emergently due to respiratory crisis. However, the increasing availability of noninvasive ventilation techniques including bilevel positive pressure support (BiPaP), inexsufflator treatments (cough assist machine), percussion, postural drainage and suction can help allow such infants to be extubated more readily than in the past, providing an intermediary between tracheostomy and withdrawal of support. Perhaps more important, however, it offers families additional options for ongoing respiratory support on a daily basis at home, thus minimizing emergency room and hospital visits, and potentially extending lifespan in more moderately affected infants.

An individualized approach to respiratory infection or compromise should be developed with the child's family and updated regularly with regard to choices surrounding a need for intubation. If the family chooses palliative care, hospice provides compassionate support for such families. The use of narcotic medication in this setting to reduce discomfort, along with proper positioning and a less aggressive respiratory support regimen, in concert with the family's wishes can minimize discomfort for the infant. It may be beneficial to have the family communicate with others who have experienced the loss of an infant with SMA type I during this difficult period.

DENTAL CARE AND TREATMENT

General Recommendations:
Children should be screened by a pediatric dentist as soon as possible following diagnosis.

Tooth Brushing:

- Minimum frequency 2x/day; an adult should brush the child's teeth from the moment the first tooth appears.
- If mechanical brushes are used, they should be used as one would a normal toothbrush, using up and down movement rather than side to side.
- Parents should check for food pouching in the cheeks and palate.
- If the child resists brushing, parents should take the approach, "I love you too much not to brush your teeth" and do it anyway.
- Poor oral hygiene increases the risk for pneumonia.

- Institute the of prevention of jaw contracture with the therabyte system at the earliest indication of tightness.
- Early referral to an Orthodontist as indicated is important to help utilize palate expanders for narrow high arched palates and teeth crowding.

Flossing:

- Flossing is every bit as important as brushing.
- Flossing should begin as soon as teeth erupt.

Diet:

- Acidic beverages are one of the most significant reasons for tooth decay.
- Absolutely no soda, energy drinks or sweetened fruit juice or beverages.
- 100% fruit juice is allowed only in small quantities (4 oz or less/day/30 lbs)
- Desired beverages include milk and water.
- Do not put a child to sleep with a bottle (if you must, please use water)
- A well balanced diet limits foods with added sucrose, fructose, sugar or related sweeteners.
- A good diet should contain calcium and vitamin D from natural sources.
- Regular dental visits for cleaning, consultations, cavity monitoring and protection such as sealants are a must.

Relevance of proactive dental hygiene and prevention of jaw contracture:
Significant jaw contractures ultimately develop in a majority of adults with type II SMA. Some children with SMA type II, and virtually all children with type I who survive to later childhood and adulthood also manifest this problem. Jaw contractures can result in inadequate oral hygiene and increase the risk for serious dental complications over time. Because of discomfort experienced during routine dental care due to this issue, many individuals choose to avoid going to the dentist at all rather than address the primary issue. This sets up a vicious cycle of poor dental care and hygiene, and ultimately, tooth decay. Subjects with tooth decay and gingivitis are at an increased risk for pneumonia as well as more serious medical problems such as endocarditis (inflammation and infection on heart valves). Thus, it is critical to avoid these problems in the first place by being proactive from early childhood. We recommend dental visits at least every six months beginning at three years of age for all individuals with SMA. Early consultation with an Orthodontist for consideration of a retainer or palate expander can help prevent jaw and facial deformity and abnormal bit alignment due to decreased use of mandibular muscles.

Anticipatory jaw stretching is one possible solution to help avoid this problem. The Therabyte System is one such solution. In subjects whom significant jaw contracture has already developed, and who require dental or oral surgery or other medical procedures requiring significant mouth opening, a gradual increase in the width of jaw opening over a period of several weeks can increase access. However, this needs to done gently and carefully to avoid damage to the temporomandibular joint and surrounding muscles. If a dental, endodontal or oral surgery is procedure is to be done, and they haven't had adequate ongoing mouth care, standard

antibiotic prophylaxis should be considered prior to such a procedure in which those who haven't had adequate preventative mouth care. Most individuals with significant jaw contracture have respiratory impairment which warrants an anesthiology consult if any type of sedation is planned. To avoid serious muscle damage and chronic temporomandibular joint pain, intubation using a laryngeal mask airway (LMA, which doesn't require visualization of vocal cords) or fiber optic intubation via placement of an endotracheal tube over a bronchoscope can ensure adequate ventilation during and after the procedure. Sometimes, use of muscle relaxants or even paralysis are necessary to enable adequate jaw-opening to extract teeth or perform dental or oral surgery procedures. Where such a procedure is performed depends on how fragile their respiratory status is. Most individuals will need to be in a place where they can get appropriate post-procedure care and support until they have recovered to their baseline state with regard to airway protection.

When dental issues have not been addressed in a proactive fashion, individuals who suffer gingivitis, tooth decay, abscesses or severe temporomandibular joint restriction can develop significant and chronic pain. While addressing the primary issue is desirable, sometimes individuals are fearful of aggressive interventions, and may need time to fully consider the risks and benefits of more invasive procedures. It is important not to overlook the importance of adequate pain control or less invasive interim solutions in such patients, including palliative care interventions.

FUTURE DIRECTIONS

Proven treatments for SMA may soon become a reality as we gain a better understanding of disease pathogenesis.

Clinical trials to assess compounds that increase SMN protein levels in cell and animal models have begun. In the meantime, a proactive management strategy can help to limit disease progression and optimize outcome.

Undoubtedly, intervening as early as possible in the disease process will prove most effective as additional therapeutic strategies are identified. Participation in clinical trials can provide patients and families with additional clinical care and monitoring that can help to ensure that proactive care is the rule, rather than the exception.

Participation is also vital to the community to help demonstrate the effectiveness of potential treatments. However, a careful consideration of the potential risks as well as the time commitment involved should be carefully considered in each case.

View a listing of clinical trials on SMA involving the University of Utah or the Project Cure SMA Investigators Network.

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Last updated 11/2007. Prepared by Mary Schroth, M.D. Pediatric Pulmonology, University of Wisconsin, and Kathryn J. Swoboda, University of Utah. Adapted from Bach JR et al, Spinal muscular atrophy type 1: a noninvasive management approach. Chest 2000;117:1100. These are guidelines only. Please consult your doctor regarding specific concerns about how these guidelines might apply to you or your child. Contact Swoboda@genetics.utah.edu for specific questions regarding this document.

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RESPIRATORY CARE OF SMA AND CHOICES

IDENTIFYING THE PROBLEM

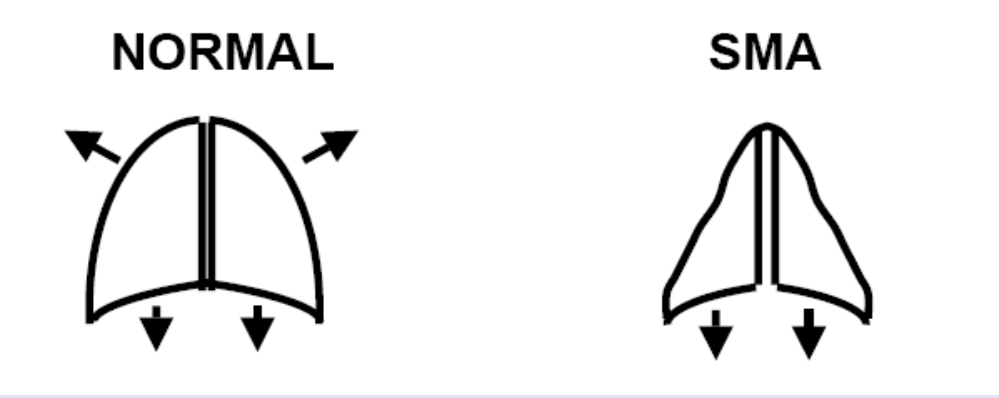
Respiratory care is critically important for individuals with spinal muscular atrophy and affects both their survival and quality of life. SMA affects the muscles for breathing as well as the muscles for moving the arms and legs.

NORMAL BREATHING MUSCLE FUNCTION

The muscles important for breathing include the intercostal muscles and the diaphragm. The intercostal muscles are the muscles between the ribs. These muscles help to lift up and expand the shape of the rib cage thereby helping the lungs to inflate. The diaphragm is the large muscle at the bottom of the rib cage. The diaphragm works by pulling the rib cage down in an almost opposite direction of the intercostal muscles, and also helps the chest wall and lungs to expand causing air to fill the lungs. What you see in someone without SMA is that the ribcage inflates with air at the top of the ribcage as well as at the bottom of the ribcage during inhalation.

SMA BREATHING MUSCLE FUNCTION

SMA causes very weak intercostals muscles, the muscles between the ribs. The strongest breathing muscle in SMA is the diaphragm. As a result, children with SMA type I and type II appear to breath with their stomach muscles. The belly or abdomen pooches out while they take a breath in. The top of the rib cage does not appear to move. In infants less than a year of age, the rib cage is very soft even though it consists of the ribs, which are bones and cartilage. Over time the rib cage may appear as though the bottom of the ribcage is pulled down. This gives the appearance of a bell shaped chest meaning that the top of the chest is narrow and the bottom of the chest is much wider. Sometimes the area of the sternum or the bone in the front that the ribs attach to becomes sunken. This is called pectus excavatum and happens because the intercostals muscles are not strong enough to help pull the top of the rib cage out against the diaphragm that is pulling down.



WHAT DOES THIS MEAN?
Individuals with SMA have respiratory muscles that are at a mechanical disadvantage. Because the lungs cannot inflate fully, the lungs may not fully develop. In addition, because the muscles are weak, the cough is very weak, and it is difficult to take deep enough breaths while sleeping to maintain normal oxygen levels and normal carbon dioxide levels.

LUNG UNDERDEVELOPMENT
Lungs that are not fully developed may contribute to decreased lung function as the child grows. Normal lung expansion helps the rib cage to grow and maintain a normal chest wall shape. A normal chest wall shape provides the best mechanical advantage for the lungs to work optimally. One of our goals for the respiratory care of children with SMA is to provide maximum lung and chest wall development.

WEAK COUGH
During viral respiratory infections, the nose and the lungs make a lot of secretions. In addition, everyone is weaker during a cold. If the secretions are not cleared from the lungs by coughing, the secretions will collect in the airways and cause plugging of the small breathing tubes of the lungs. If the secretions become infected this will result in pneumonia. Plugging of the small breathing tubes of the lungs results in collapse of some of the air sacs and this is called atelectasis. A consequence of atelectasis is decreased oxygen saturation, which means that there is not enough oxygen in the blood for the body to maintain normal functions. Oxygen saturation can be determined easily using a pulse oximeter.

SHALLOW BREATHING DURING SLEEP
During sleep, the muscles relax including the muscles for breathing. During normal breathing, we breathe in oxygen and exhale carbon dioxide. When someone with muscle weakness falls asleep, their muscles for breathing also relax and their breaths become more shallow. Sometimes those breaths become so shallow that they do not inhale adequate oxygen and they also do not exhale enough carbon dioxide. This is called hypoventilation. As a result, their oxygen saturations during sleep may drop to less than 94% and their carbon dioxide level may increase.

WHAT CAN BE DONE

LUNG DEVELOPMENT
Lung underdevelopment can be overcome by giving insufflations or big breaths daily using any of the following: a resuscitator bag and mask, or a cough machine on the inspiratory setting only or an intermittent positive pressure breathing (IPPB) device. This exercises the muscles for breathing and expands the lungs to a larger volume than the person can do on their own.

1. Using the **resuscitator bag and mask**, the mask is placed over the nose and mouth and a breath is given to the person while they are

inhaling. Several breaths are given over several minutes to help inflate the lungs.

2. A **cough machine** with an automatic cycle is a device that can be set to shift from
inhalation mode to exhalation mode and then a pause before the next breath. The breaths are given by either a mask over the nose and mouth or a mouthpiece. Using the cough machine in the inhalation mode only, the lungs can be inflated to a set pressure. (Please see additional information below in Improve Cough.)

3. **IPPB** gives a breath in synchrony with an individual to a set pressure with a mouthpiece or a mask over the nose and mouth. A similar effect can be achieved using the cough machine.Incentive spirometers typically given to adults after a surgery are not helpful to individuals with SMA because the spirometer inflates the lungs but only to the volume the person can do already. There is no extra expansion or benefit to using this device in individuals with SMA type I and II.

IMPROVE COUGH/AIRWAY CLEARANCE

Coughing up secretions can be improved dramatically by using a cough machine or having someone else use their hand to assist with coughing. Manual cough assist means that another person uses their hand to apply pressure under the diaphragm in synchrony with the coughing person. This results in increased force of the diaphragm to move air out of the lungs.

The cough machine inflates the lung to a set pressure for a set time following by sucking the air out of the lungs at a set pressure for a set time. This is very similar to what occurs during coughing in stronger people. When you cough, you inhale a big breath, close your throat, increase the pressure inside your chest, open your throat and then the air bursts out of the lungs.

Typical settings for the cough machine are inhale pressure 30 to 40 cm of H20 for 1 second, exhale pressure –30 to –40 for 1 second and pause time 1 to 2 seconds. Four to 5 breaths are taken continually followed by suctioning the mouth to remove secretions coughed up and a rest period of 1 to 2 minutes. Four sets of 5 breaths is a typical treatment.

The cough machine cannot be overused for the purpose of coughing and should be thought of as the way the person with SMA coughs. There is no other device that helps to clear the secretions out of the lungs. The cough machine is available through your home care durable medical equipment (DME) provider. The cough machines are known by brand as the In-exsufflator Cough Machine or the newer Cough Assist.

AIRWAY SECRETION MOBILIZATION

When a person with SMA has a cold, the secretions build up and are difficult to cough out. One of the ways we help remove secretions is to loosen them in the airways before coughing. The techniques available to loosen secretions include manual or mechanical percussion, postural drainage, intrapulmonary percussive ventilation (IPV), and the Vest Airway Clearance system. No one technique has been proven to be better than any other; they all mobilize secretions.

1. Manual or mechanical percussion means using your hands or palm cups to clap on different parts of the chest while positioning the person to optimally drain that part of the lung. Mechanical percussion refers to using a vibrating device applied to the chest to loosen secretions.

2. Postural drainage means positioning the person so that the secretions in the lungs will drain with gravity. Typically the person will lay on their tummy, back or side with their bottom higher than their chest. This allows the secretions to move with gravity from the smaller airways to the larger airways.

3. IPV is a mechanical device that provides positive pressure by mask or mouthpiece into the airways at a set frequency and pressure that can be varied as tolerated. Medication or saline is nebulized with IPV. This is mechanical percussion from the inside of the lungs. The machine sounds like a choo-choo train.

4. The Vest Airway Clearance System is a mechanical device that delivers air at a set frequency and pressure to the chest through a vest that the individual wears. This device is expensive and has not been proven to be more effective than other modes of airway secretion mobilization.

BREATHING SUPPORT OPTIONS

Individuals with SMA type I and many individuals with SMA type II have hypoventilation while they sleep at night. This means that they exchange carbon dioxide and oxygen at lower levels than is necessary for the body to maintain normal function. Their oxygen saturations drop and their carbon dioxide levels increase. To improve this problem, patients with SMA have to receive bigger breaths during sleep than they can do on their own. There are several options to accomplish better ventilation (bigger breaths) and oxygenation while sleeping including BiPAP, mechanical ventilator, and negative pressure ventilation. During illnesses the respiratory support device may need to be worn throughout the day also. **There is no role for supplemental oxygen in patients with SMA** until **ALL** other options have been exhausted.

1. BiPAP refers to bilevel or two level positive airway pressure. This is usually delivered through a mask over the nose. BiPAP provides a higher pressure and volume of air into the lungs during inhalation and inflates the lung greater than what the person can do without the

BiPAP machine. During exhalation, the BiPAP pressure drops so that air can passively leave the lungs. The BiPAP machine can sense when the person is taking a breath and gives the breath in synchrony with the individual. A respiratory rate is also set so that the BiPAP will give breaths even when an individual with SMA is in a deep sleep and taking shallow breaths. The person can breath above the BiPAP rate and the BiPAP will deliver more breaths. CPAP (continuous positive airway pressure) should never be used in patients with SMA. Typical BiPAP settings in an individual with SMA include are inspiratory positive airway pressure (IPAP) 14-20, expiratory positive airway pressure (EPAP) 3-6, respiratory rate based on age of 14-30, inspiratory time based on age of 0.5 to 1.5 seconds, and respiratory rate and rise time based on comfort.

2. Mechanical ventilators or respirators come in a variety of models. The most portable model that is available is the LTV (laptop ventilator). This ventilator can be carried over the shoulder or placed on a wheelchair for transport. Portable external batteries are also available. Mechanical ventilators are more complex, but also allow for control of more variables. The ventilator can be set to deliver a specific size breath and a set number of breaths per minute. Mechanical ventilation can be delivered with a nose mask, mouthpiece while awake, or through a tracheostomy tube.

3. Negative pressure ventilation refers to providing breaths into the lungs using a large chamber or tank that encircles the chest. The chamber is connected to a vacuum pump that takes the air out of the chamber and as a result the chest wall expands to bring air into the lungs. A Porta-Lung is an example of a negative pressure ventilator. It can be set to deliver a specific number of breaths per minute and a vacuum pressure.

PROTOCOL FOR MANAGING AIRWAY SECRETIONS DURING A COLD

During a cold, individuals with muscle weakness can have a very hard time clearing mucus from their lower airway. You can help them by following these steps:

1. These steps should be done in the order shown below every **four hours** and as needed during the cold.
    a. **Airway secretion mobilizat**ion for 10-20 minutes.
    b. **Cough machine**, four sets of 5 breaths to remove the loosened secretions followed by suctioning secretions from the mouth.
    c. **Postural drainage** for 15-30 minutes, which means laying with their bottom higher than their chest so that gravity helps to move the secretions out of the lungs.

d. **Cough machine**, four sets of 5 breaths to help remove the remaining drained secretions followed by suctioning secretions from the mouth.

2. Use the cough machine every time your child sounds rattley or has trouble coughing out secretions. You cannot use this machine too much. If your child has difficulty getting secretions out of the back of their throat, also suction their mouth.

3. If your child is on BiPAP or breathing support by nose mask, use the machine every time that they are asleep including naps during the day. Your child is weaker than usual during colds. They may also need to use the breathing support machine while they are awake.

4. Use a pulse oximeter to check your child's oxygen saturations as needed when well and more frequently when sick. Often you may not be able to tell when your child is having difficulty breathing and the pulse oximeter may tell you there is a problem sooner and that they need help coughing. If the oxygen saturation is less than 94%, use the cough machine to clear secretions and recheck the oximetry. If the oxygen saturation is 92% or less and the cough machine is not increasing the oxygen saturation, place your child on BiPAP or equivalent and give airway clearance treatment and please contact your physician. If you cannot maintain oxygen saturation greater than 90% while on BiPAP and in room air, your child may need to be in the hospital for more intense therapy.

5. If your child is vomiting and not tolerating nutrition, they may become dehydrated. This can be a very serious complication in children with Spinal Muscular Atrophy. Please contact your physician.

CHOICES

Non-invasive respiratory care management means helping a child to breathe using a mask over the nose that is connected to a BiPAP machine or a mechanical ventilator. Invasive respiratory management refers to the use of a breathing tube that is placed in the mouth and passed between the vocal cords and into the lungs. Invasive respiratory management also includes the placement of a surgical hole in the neck to the large airway (trachea) for a tracheostomy tube. A tracheostomy tube bypasses the mouth and vocal cords and goes directly from the skin to the trachea. A respirator or ventilator is connected to the endotracheal tube or tracheostomy tube.

SMA TYPE I

Children with SMA type I have a huge variation in muscle strength and their breathing support needs are difficult to predict. Almost all children with SMA type I benefit from supportive ventilation with BiPAP or a ventilator using a nasal mask

while they sleep starting at an early age, 6 months for example. The breathing support needed while healthy ranges from no breathing support while sleeping to needing BiPAP while sleeping to needing breathing support 24 hours per day, 7 days per week with or without a tracheostomy tube. During colds all children require additional breathing support and may need to use their BiPAP or ventilator 24 hours per day until the cold goes away.

Parents of children with SMA type I have many very difficult decisions. First is whether you wish to pursue non-invasive breathing support per nose mask for your child. The methods and equipment although generally effective are also a tremendous amount of work, especially when the child has a cold. In addition, the child may not tolerate the nose mask breathing support. Another decision is whether you desire to have your child intubated should they become very sick especially with a cold. Remember, everyone becomes weaker with a cold.

One of the things you and your doctor should consider is whether the illness and difficulty breathing is a sudden change due to a cold and may be reversible, or whether it seems to be a result of the child becoming gradually weaker over all. Using an aggressive airway clearance and extubation (removal of the breathing tube) protocol followed by placing the child on breathing support by nose mask provides the greatest chance for success.

Some physicians are not aware of the protocol and tools that are available. However, when a child with SMA type I has a breathing tube placed one of the risks is whether the child will succeed in having the breathing tube removed and using breathing support by nose mask when the illness has resolved. If the child does not tolerate breathing support by nose mask despite the best efforts of the doctor another difficult decision has to be made.

The decision at this point is whether to:

1. Wait and see if the child will become stronger and try again.
2. Consider transferring to a hospital that has a lot of experience with non-invasive respiratory care if your hospital does not.
3. Consider tracheostomy tube placement.
4. Consider letting your child go.

There are no right answers and as a parent, you will make the best decision that you can for your child. Something to keep in mind is that the non-invasive methods for respiratory care of children with SMA are not for everyone and not everyone can do this for a variety of reasons. In addition, some children do not do well with the non-invasive breathing support or require the breathing support 24 hours per day. Requiring 24 hours per day breathing support by nose mask may be very difficult for the child as well as for the family. Again, you as the parent will make the best decision you can for your child.

SMA Type II

Children with SMA type II should never require a tracheostomy, and in general are strong enough to be managed using non-invasive respiratory care techniques on a

day to day basis. This means that when the child is healthy, they may use BiPAP or a ventilator with a nose mask or negative pressure ventilation while they sleep, and the Cough Assist device as needed. Some children with SMA type II do not require support for their breathing while they sleep.

When a child with SMA type II develops a cold, they may need to use their nose mask breathing support more often and sometimes continuously. Remember that during a cold they are weaker than usual, but their strength improves after the cold has resolved. During a cold they 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. If oxygen saturation cannot be maintained at 92% or greater on BiPAP, they may need to be hospitalized and may need to have a breathing tube place and attached to a ventilator. However, through aggressive use of airway secretion mobilization and airway clearance, the tube can be removed and the child placed on ventilation by nose mask and then weaned back to using the breathing support while they sleep only.

SUMMARY

The respiratory care for children with SMA is critically important. The necessary tools include a cough machine, a method for airway secretion mobilization, a method for performing postural drainage, and a pulse oximeter to help monitor their oxygen saturations. In addition, a method for breathing support including a BiPAP machine or mechanical ventilator are also very important. For individuals with SMA type II, aggressive respiratory care using non-invasive techniques improves quality and quantity of life. For children with SMA type I, the non-invasive respiratory care techniques are beneficial and generally effective. Please keep in mind that not all techniques fit every child.

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SPINAL MUSCULAR ATROPHY: CHOICES

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Respiratory Care of SMA and Choices
Revised 030908

Last updated 11/20/07
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Adapted from Bach JR et al, Spinal muscular atrophy type 1: a noninvasive respiratory management approach. Chest 2000; 117:1100. These recommendations are guidelines only; please consult with your physician regarding specific care issues in you or your child. For questions regarding this document, you may email Swoboda@genetics.utah.edu.

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NON-INVASIVE MECHANICAL VENTILATION FOR SMA TREATMENT

EXPIRATORY AIDS

Inspiratory and expiratory muscle aids are devices and techniques that involve the manual or mechanical application of forces to the body or intermittent pressure changes to the airway to assist inspiratory or expiratory muscle function. The most important inspiratory aid is to receive air under pressure when one inhales (intermittent positive pressure ventilation or IPPV). The most important expiratory aid is to have a negative pressure (vacuum) applied to the airway via the nose and mouth when one coughs along with a manual thrust to the belly to further increase cough flows.

No one should receive supplemental oxygen, bronchodilators, or other medications as an alternative to normalizing blood oxygen levels by normalizing lung ventilation. Using oxygen rather than assisted ventilation results in worsening of carbon dioxide retention and inevitably results in respiratory failure.

Manually Assisted Coughing

Illness and death in people with generalized weakness is almost always due to respiratory difficulty that occurs because of a weak cough. Breathing (inspiratory), expiratory, and throat (bulbar) muscles are needed for effective coughing. The latter are predominantly the abdominal muscles. Clearing airway secretions can be a continual problem but it most often occurs during chest infections and following general anesthesia for surgery for any reason.

Peak cough flows (PCF) most often exceed 160 liters per minute to be effective for coughing up airway debris. PCF are increased by manually assisted coughing. If the vital capacity is less than 1.5 liters, insufflating or air stacking to the maximum insufflation capacity (MIC) becomes crucial to optimize cough flows. Once the person is insufflated to the MIC, an abdominal thrust is timed to the cough to increase the flow. Techniques of manually assisted coughing involve different hand and arm placements for thrusts. A belly thrust with one hand while applying counterpressure to the chest with the other arm and hand further increases assisted PCF for 20% of people.

Manually assisted coughing requires a cooperative patient, good coordination between the patient and caregiver, and adequate physical effort and often frequent application by the caregiver. It is usually ineffective in the presence of significant back deformity. Abdominal compressions should not be used for one and one-half hours following a meal, however, chest compressions can be used to augment PCF. The inability to generate over 160 liters per minute of assisted PCF despite having a

vital capacity or MIC greater than 1 liter usually indicates fixed upper airway obstruction or severe throat muscle weakness and airway collapse during coughing attempts. Vocal cord adhesions or paralysis may have resulted from a previous translaryngeal intubation or tracheostomy. Some lesions, especially the presence of obstructing granulation tissue, can be corrected surgically. When inadequate, the most effective alternative for generating optimal PCF and clearing airway secretions is the use of MI-E.

Mechanical Insufflation-Exsufflation (MI-E)

Mechanical Cough Assist devices (J. H. Emerson Co., Cambridge, MA) deliver deep insufflations followed immediately by deep exsufflations. The insufflation and exsufflation pressures and delivery times are independently adjustable. Except after a meal, an abdominal thrust is applied in conjunction with the exsufflation. MI-E can be provided via an oral-nasal interface, a simple mouthpiece, or via an invasive airway tube like a tracheostomy tube. When delivered via the latter, the cuff, when present, should be inflated.

The Cough-Assist can be manually or automatically cycled. Manual cycling facilitates caregiver-user coordination of inspiration and expiration with insufflation and exsufflation, but it requires hands to deliver an abdominal thrust, to hold the mask on the patient, and to cycle the machine. One treatment consists of about five cycles of MI-E followed by a short period of normal breathing or ventilator use to avoid hyperventilation. Insufflation and exsufflation pressures are almost always from +35 to +60 cm H2O to -35 to -60 cm H2O. Most patients use 35 to 45 cm H2O pressures insufflations and exsufflations. In experimental models, +40 to -40 cm H2O pressures have been shown to provide maximum forced deflation volumes and flows (see (3)further information). Multiple treatments are given in one sitting until no further secretions are expulsed and any secretion or mucus induced oxygen desaturations are reversed. Use can be required as frequently as every few minutes around the clock during chest colds. Although no medications are usually required for effective MI-E in people with weak muscles, liquefaction of sputum using heated aerosol treatments may facilitate exsufflation when secretions are inspissated.

Whether used via the nose or mouth or via invasive indwelling airway tubes, routine airway suctioning misses the left lung about 90% of the time so that 80% of pneumonias are in the left lung. MI-E via an airway tube provides the same exsufflation flows in both left and right airways without the discomfort or airway trauma of tracheal suctioning and it can be effective when suctioning isn't. Patients invariably prefer MI-E to suctioning for comfort and effectiveness and they find it less tiring. Deep suctioning, whether via airway tube or via the upper airway, can essentially be discontinued for most patients.

The use of MI-E has permitted us to consistently extubate people with neuromuscular disease following general anesthesia despite their lack of any ability to breathe on their own, and to manage them with noninvasive IPPV. It has also permitted us to avoid intubation or to quickly extubate people in acute ventilatory failure and with profuse airway secretions due to intercurrent chest infections. MI-E

in a protocol with manually assisted coughing, oximetry feedback, and home use of noninvasive IPPV was shown to effectively decrease hospitalizations and respiratory complications and mortality for people with neuromuscular diseases. It may not be effective if the user can not cooperate sufficiently to keep the airway open, if there is a fixed upper airway obstruction, or if upper airway dilator muscles can not maintain sufficient patency to allow for PCF to exceed 160 L/m. This is most often seen in advanced amyotrophic lateral sclerosis with severe throat muscle weakness. MI-E has been demonstrated to be extremely safe even when used many times at pressures of 40 to 60 cm H2O. It is rarely effective for people of any age at pressures less than 35 cm H2O.

INSPIRATORY AIDS

Inspiratory and expiratory muscle aids are devices and techniques that involve the manual or mechanical application of forces to the body or intermittent pressure changes to the airway to assist inspiratory or expiratory muscle function. The most important inspiratory aid is to receive air under pressure when one inhales (intermittent positive pressure ventilation or IPPV). The most important expiratory aid is to have a negative pressure (vacuum) applied to the airway via the nose and mouth when one coughs along with a manual thrust to the belly to further increase cough flows.

No one should receive supplemental oxygen, bronchodilators, or other medications as an alternative to normalizing blood oxygen levels by normalizing lung ventilation. Using oxygen rather than assisted ventilation results in worsening of carbon dioxide retention and inevitably results in respiratory failure.

Maintaining Lung Range-of-Motion
Lungs become stiff if not expanded regularly. The vital capacity is the deepest breath that one can blow into a measuring device called a spirometer. If the vital capacity is 500 ml but the predicted capacity is 5000 ml then without assistance one can only expand about 10% of one's lungs and the rest closes down and for children does not grow properly. Use of incentive spirometry or deep breathing is useless because it does not expand the lungs more than about 10%. Mobilization of the lungs to prevent chest wall contractures and lung restriction can only be achieved by providing regular deep volumes of air (insufflations) or overnight deep breaths (IPPV).

A person's maximum insufflation capacity (MIC) is determined by giving the person the largest volume of air that he or she can hold with the throat closed. This is usually done by teaching the person to stack volumes of air consecutively delivered from a manual resuscitator. The person holds the stacked air with the throat (glottis) closed until no more air can be held. Patients who learn glossopharyngeal (frog) breathing can often air stack to the MIC without mechanical assistance.
The primary objectives in using air stacking or in providing maximum insufflations as lung and chest wall range-of-motion are to: increase the MIC, to maximize

cough flows, to maintain or improve lung elasticity, to prevent or eliminate atelectasis, and to master noninvasive ventilation (noninvasive IPPV). The ability to air stack means that one can use noninvasive ventilation and assisted coughing to prevent pneumonia, respiratory failure, or the need to ever undergo tracheostomy. Air stacking can also increase voice volume, facilitate eating, and promote lung growth in children.

Since anyone who can air stack is also able to use noninvasive IPPV, if such a patient is intubated for respiratory failure, he or she can be extubated directly to continuous noninvasive IPPV whether or not able to breathe independently. (see intensive care protocols).

Noninvasive IPPV

IPPV can be noninvasively delivered via mouth pieces, nasal, and oral-nasal interfaces for nocturnal ventilatory assistance . Many people with no ability to breathe on their own and no upper limb function keep simple Respironics (Murrysville, PA) 15 mm or 22 mm angled mouth pieces near their mouths and grab them between their teeth as needed to receive mouth piece IPPV during daytime hours. Mouth piece IPPV is the most important method of daytime ventilatory support. Some people keep the mouth piece between their teeth all day. Most prefer to have the mouth piece held near the mouth. A metal clamp attached to a wheelchair can be used for this purpose or the mouth piece can be fixed onto motorized wheelchair controls, most often, sip and puff, chin, or tongue controls. The ventilator is set for much greater than normal tidal volumes, often from 1000 to 2000 ml. The person grabs the mouth piece with his mouth and supplements or substitutes for inadequate breath volumes. The person varies the volume of air taken from ventilator cycle to ventilator cycle and breath to breath to vary speech volume and cough flows as well as to air stack to fully expand the lungs.

To use mouth piece IPPV effectively and conveniently, adequate neck rotation and oral motor function are necessary to grab the mouth piece and receive IPPV without insufflation leakage. Since the low pressure alarms of volume-cycled ventilators can often not be turned off, to prevent their sounding during routine daytime IPPV when not every delivered volume is received by the patient, a flexed mouth piece for IPPV or an in-line regenerative humidifier can be used. These create 2 or 3 cm H2O back pressure which is adequate to prevent the low pressure alarm from sounding.
The lipseal can provide an essentially closed system of noninvasive ventilatory support when using mouth piece IPPV during sleep. Lipseal IPPV is delivered during sleep with little loss of air out of the mouth and with virtually no risk of the mouth piece falling out of the mouth. Orthodontic bite plates and custom fabricated acrylic lipseals can also increase comfort and effectiveness. Typically high ventilator insufflation volumes of 1000 to 2000 ml compensate for air leakage out of the nose during sleep.

Because people prefer to use mouth piece IPPV or the intermittent abdominal pressure ventilator for daytime use, nasal IPPV (or the noninvasive delivery of IPPV via a nasal interface "CPAP mask") is most practical only for nocturnal use. Daytime nasal IPPV is indicated for those who can not grab or retain a mouth piece because

of oral muscle weakness, inadequate jaw opening, or insufficient neck movement. Twenty-four hour nasal IPPV can, nevertheless, be a viable and desirable alternative to tracheostomy even for some people with severe lip and mouth muscle weakness.

Most people prefer to use IPPV via a nasal rather than oral interface during sleep. Whether using nocturnal nasal or lipseal IPPV in a regimen of 24 hour noninvasive IPPV, and despite the maintenance of normal daytime alveolar ventilation, about 3% of people with no ability to breathe on their own have episodes of excessive air loss during sleep. These often result in arousals with shortness of breath. The person may also complain of recurrence of morning headaches, fatigue, and perhaps nightmares and anxiety. The nasal ventilation user should usually be switched to lipseal IPPV and lipseal IPPV users can have their systems "closed" by having their nostrils clipped or plugged with cotton kept in by covering the nostrils by a bandaid during sleep. Another practical solution is to set the ventilator's low pressure alarm at a level that, by its sounding, stimulates the sleeper sufficiently to shorten periods of air leaking during sleep. Commonly, a low pressure alarm setting of 10 to 20 cm H2O pressure is used for this purpose and the user develops sleep reflexes to prevent prolonged air leakage.

There are now numerous commercially available nasal interfaces (CPAP masks). These include the Monark and gel masks from Respironics Inc., Murrysville, CO, the ResCare Inc. (San Diego) Sullivan mask, the SleepNet (Manchester, N.H.) Phantomtm and IQtm Nasal Masks, and Mallincrodt interfaces (Pleasanton, CA). Each interface design applies pressure differently to the paranasal area. One can not predict which model will be most effective and preferred by any particular user. Skin pressure and insufflation leakage into the eyes are common complaints with several of these generic models. Such difficulties resulted in the fabrication of interfaces that mold themselves to facial tissues and of other custom molded interface designs . People must be offered trials using various nasal interfaces and are encouraged to choose between them. Interface use is evaluated for comfort and seal around the nose. No one should be offered and expected to use only one nasal interface anymore. Alternating IPPV interfaces nightly alternates skin pressure sites, minimizes discomfort, and is to be encouraged.

People whose blood carbon dioxide levels increasing during the day causing their blood oxygen levels to decrease below 95% need to use noninvasive IPPV, usually mouth piece IPPV, for periods of time during daytime hours. Failure to maintain normal lung ventilation during daytime hours will result in inadequate nocturnal benefit from the use of noninvasive IPPV. For patients not wishing to switch to lipseal IPPV for nocturnal aid despite excessive air leakage out of the mouth, a chin strap or plugged lipseal can be used to decrease mouth leakage. In the presence of nasal congestion people either use decongestants to permit nasal IPPV or they switch to mouth piece and lipseal IPPV, or on rare occasions, temporary use of a body ventilator like a portable iron lung. Most often the person continues nasal IPPV using decongestants.

Because of the need for air stacking, people over 5 years old whose MICs exceed their vital capacities need to use volume cycled portable ventilators rather than pressure cycled machines like BiPAP because the latter can not provide optimally deep breaths or permit users to stack breaths.

In summary, noninvasive IPPV can be used for up to full-time ventilatory support for the great majority of people with no ability to breathe on their own provided that they have mouth muscle function sufficient for speaking.

OUTPATIENT CONTROL

Patients are considered to be at risk for respiratory failure particularly during chest colds when they have assisted PCF below 270 liters per minute. They are prescribed oximeters and trained in air stacking insufflated volumes via mouth and nasal interfaces, manually assisted coughing, MI-E at +35 to +50 to -35 to -50 cm H2O pressure drops with abdominal thrusts applied during exsufflations, and given rapid (less than 1 hour) access to a portable volume ventilator, a Cough Assist (J. H. Emerson Co., Cambridge, MA), and to various mouth pieces and nasal interfaces.

The patients and care providers are instructed that any decreases in SaO2 below 95% indicate either hypoventilation or the presence of airway mucus accumulation that must be cleared to prevent atelectasis and pneumonia. They are told to use oxyhemoglobin saturation (SaO2) monitoring whenever fatigued, short of breath, or ill. They use noninvasive IPPV and manually and mechanically assisted coughing as needed to maintain normal SaO2 at all times.

Patients with elevated end tidal blood carbon dioxide levels or periods of daytime SaO2 below 95% undergo nocturnal SaO2 monitoring. When symptomatic or nocturnal means are below 94% a trial of nocturnal nasal IPPV is provided. People continue to use nocturnal nasal IPPV when they felt less fatigue and nocturnal mean SaO2 increases. Most young patients use noninvasive IPPV for the first time to assist lung ventilation during chest infections.

TECHNIQUES FOR CHILDREN

The parents of infants with neuromuscular disorders, most often, infants with SMA types 1 and 2, are told that their children will not survive infancy. Typically, these children are well until a cold causes airway secretions to accumulate in the lungs because of a weak cough mechanism. Bacteria multiply in the secretions and the child develops pneumonia and respiratory failure. After hospital admission, the child is typically intubated (a tube is passed via the nose or mouth into the lungs) for ventilatory support and airway suctioning. Attempts to remove the tube usually fail because the methods used to do so are appropriate for children whose primary disorder is lung disease rather than muscle weakness. Once extubation has failed,

the parents are told to either let the child die or to agree to tracheotomy (placing a tube permanently in the neck for ventilatory support and suctioning). We have found that this is usually unnecessary when noninvasive inspiratory and expiratory muscle aids are used appropriately.

Infants cannot air stack delivered volumes of air or cooperate to receive maximal insufflations. All SMA type 1 babies, SMA type 2 infants and others with infantile neuromuscular weakness who have paradoxical chest wall movement (inward movement of the chest when the belly goes outward during inspiration), require nocturnal high span bi-level positive airway pressure to prevent pectus excavatum (chest deformity) and promote lung growth. (See article 143 Dr. Bach's Bibliography). Nocturnal high span bi-level positive airway pressure prevents or reverses pectus excavatum. It should be introduced to the small child once he or she is asleep. The child gradually becomes accustomed to it. Inspiratory pressures should begin at 15 cm H2O and be gradually increased to 18 to 20 cm H2O.
Expiratory pressures should be left at the minimum or 2 cm H2O. In addition to nocturnal aid, deep insufflations are provided via oral-nasal interface for children over 1 year of age along with concomitant abdominal thrusts to prevent abdominal expansion so as to direct the air into the upper chest. These insufflations need to be timed to the child's inspirations. Children can become cooperative with deep insufflation therapy by 14 to 30 months of age.

We have routinely used MI-E via translaryngeal and tracheostomy tubes in children with SMA under 1 year of age. Although we have been using approximately the same pressure settings as for adults, that is, +40 to -40 cm H2O, its use through the very narrow gauge pediatric tubes results in a severe pressure drop off that may not permit the generation of optimal exsufflation flows. However, even at these probably inadequate pressures secretions are expulsed and dSaO2s reversed. The use of MI-E via the upper airway can be effective for children as young as 13 months of age. Children this young can become accustomed to MI-E and permit its effective use by not crying or closing their throats. Between two and one-half and five years of age most children become able to cooperate and cough on cue with MI-E and they can then avoid hospitalizations and pneumonias during their intercurrent chest infections. Exsufflation timed abdominal thrusts are also used for infants.

Children too small to cooperate with manually and mechanically assisted coughing or those who are not introduced to these methods before a cold develops into pneumonia and respiratory failure are hospitalized and usually intubated (an catheter is passed via the nose or mouth into the lungs for ventilatory support and airway secretion evacuation). Often, each time that small children develop chest infections until they are old enough to cooperate will need to be hospitalized and intubated. Our intensive care protocol must be used to return these children home without the need to do a surgical tracheostomy. Patients using these methods as outpatients can also avoid respiratory complications and hospitalizations (see outpatient protocol).

NUTRITION: THE AMINOACID DIET (AA DIET)

SMA is a disorder of muscle and of fatty acid metabolism. Within 3 hours of a normal meal, blood amino acid levels of children with SMA decrease to levels that would not be reached until after at least 8 hours of fasting in normal children. In addition, infants with SMA type 1 do not efficiently metabolize fatty acids, a major source of energy during fasting. These children can have high levels of fatty acid by products in their urine and blood after overnight fasting.

In addition to metabolic aberration associated with immobility, systemic illness, muscle denervation, and muscle atrophy, SMA patients have inborn metabolic abnormalities in mitochondrial fatty acid oxidation and carnitine metabolism. Any process that increases cytoplasmic free fatty acid levels, such as fasting or defects of fatty acid transport or betaoxidation, would be expected to increase the liver and kidney's production and excretion of dicarboxylic acids. Fasting ketosis reflects normal ketogenesis by utilization of free fatty acids by the livers of SMA patients, but defective beta-oxidation of fatty acids by muscle causes fatty acid metabolites like dicarboxylic acids to spill into the blood. Dicarboxylic acid levels are elevated in the urine of infants with SMA type 1 patients tolerate the briefest fasting without ketosis and dicarboxylic aciduria whereas SMA type 3 patients express these abnormalities only during prolonged fasting, illness and periods of physiologic stress.

With relatively minor fasting infantile SMA patients develop dicarboxylic aciduria similarly to patients with primary defects of mitochondrial fatty acid beta-oxidation. (1) Metabolic analyses including the appearance of relatively early ketosis and selective renal loss of carnitine (2) and fatty vacuolization of the liver, suggest that the abnormalities are caused by changes in the cellular physiology related to the molecular defects of the SMA pathogenic Survival Motor Neuron gene or neighboring genes. Thus, the defect may be epigenic to the molecular pathogenesis of SMA itself or related to another function of the primary genetic defect. It may also contribute to the development of SMA. (1) Abnormal fatty acid metabolism also appears to resolve with age independent of disease severity.

(1) Very often before 10 years of age or during periods of physiologic stress, SMA patients suddenly lose muscle strength at a high rate. (3) Loss of strength is often triggered by respiratory tract infections and other episodes of physiologic stress and under nutrition and tends to become progressive and is most severe in infants. It is very likely that the muscle weakening in SMA infants is due to the inborn errors in fatty acid oxidation rather than to primary SMA denervation and that the weakening can be abated or averted with dietary manipulation. Diets high in carbohydrate, amino acids and polypeptides, and low in fat provide muscle with utilizable energy substrates thereby decreasing dependence on fatty acid oxidation and decreasing excessive accumulation of potentially toxic free fatty acids which can further damage muscle.

This diet maintains more normal blood glucose levels during fasting, delays fasting associated ketoacidosis, and has been noted to normalize liver function enzyme

levels. Provision of amino acids and short chained peptides instead of complex dairy proteins facilitates glucogenesis and also appears to have a beneficial effect on decreasing airway secretion production for some children.

This modified diet provide high carbohydrate and elemental amino acids and small chained polypeptides. Tolerex and Pediatric Vivonex (Novartis, Minneapolis) are two AA formulas.

WHO CAN BE HELPED at UMDNJ NON-INVASIVE RESPIRATORY CENTER

All people reach a physiological peak at age 19. Muscle strength and endurance peak around this time or soon afterwards. After this we lose 1 to 3% of our strength per year. People, such as those with post-poliomyelitis or spinal cord injury or with neuromuscular diseases such as Duchenne muscular dystrophy, spinal muscular atrophy, amyotrophic lateral sclerosis or other conditions, speak with less strength and have difficulties at an earlier age than the rest of us. The difficulties encountered from muscular weakness are in day to day functioning and in breathing and coughing due to weakness of inspiratory and expiratory (respiratory) muscles. Even in "normal" elderly people in nursing care facilities complications related to respiratory muscle weakness are a leading cause of suffering and death (see article 65 of Dr. Bach's Bibliography)

People with generalized weakness can have difficulty functioning that can be abetted with the use of assistive equipment and training. These conditions can also be a threat to survival when breathing, coughing, throat, and heart muscles are very weakened. Fortunately, today, much can be done to avoid suffering and death from these causes. Typically, muscular conditions as opposed to neurological conditions do not affect the heart muscle. However, all people with myopathies like Duchenne muscular dystrophy should be evaluated for heart muscle weakness regularly. This is particularly true today because we have new medications and medical approaches to improving heart function (see article 135 of Dr. Bach's Bibliography).

Typically people with weak breathing and coughing muscles can function appropriately until a simple cold causes the production of airway secretions that the person is too weak to cough out. Bacteria multiply in the secretions and the person develops pneumonia and respiratory distress. He or she is taken to a local hospital where oxygen is given and when breathing is suppressed by the delivered oxygen, a tube is placed through the nose or mouth into the lungs to evacuate the secretions and provide breathing support. While receiving support through the invasive tube, breathing on one's own may be delayed or thought to be impossible. The patient is then told that a tracheostomy tube is needed. This is a tube that is passed through the neck and into the airway. Typically, the hospitalization during which a person undergoes tracheostomy following pneumonia is 72 days. In reality, virtually no one who can speak needs or should ever receive a tracheostomy tube and those who have them should consider having them removed, whether needing

to use a ventilator or not. After tube removal the great majority of ventilator users can wean off of supported ventilation whereas this rarely happens when one has a tracheostomy tube (see article 76 of Dr. Bach's Bibliography).

Unfortunately, people are left to develop pneumonia after pneumonia, have hospitalization after hospitalization, and develop breathing difficulties and they are not offered the respiratory muscle aids that can prevent these problems. All that is typically done is that patients are given oxygen that only makes the problem worse and told that they must undergo tracheostomy to survive. Yet, all people with experience using tracheostomy tubes and noninvasive inspiratory and expiratory muscle aids greatly prefer the latter (see article 15 of Dr. Bach's Bibliography).

Patients helped by the Center include those with the following disorders:

- Myopathies
- Muscular dystrophies

Dystrophinopathies--Duchenne, Becker, limb-girdle, Emery-Dreifuss, facioscapulohumeral, congenital, childhood autosomal recessive, and myotonic dystrophy.

- Non-Duchenne myopathies

congenital and metabolic myopathies like acid maltase deficiency; inflammatory myopathies such as polymyositis; diseases of the myoneural junction such as myasthenia gravis; mixed connective tissue disease; myopathies associated with systemic conditions such as with cancer, cachexia/anorexia nervosa, and medications.

- Neurological disorders

spinal muscular atrophies; motor neuron diseases; poliomyelitis; neuropathies; hereditary neuropathies; any conditions with diaphragm paralysis; Guillain-Barr Syndrome; multiple sclerosis; Friedreich's ataxia; spinal cord lesions of any cause; sleep breathing disorders; familial dysautonomia; Downs syndrome.

- Skeletal and Connective Tissue Disorders

kyphoscoliosis; osteogenesis imperfecta; rigid spine syndrome; spondyloepiphyseal dysplasia congenita.

- Restrictive Lung Disease associated with lung resection, tuberculosis, Milroy's disease
- Chronic Obstructive Pulmonary Disease

INTENSIVE CARE PROTOCOL

Conventional care of patients in respiratory distress most often leads to permanent tracheostomy. Typically, the following steps are taken in the intensive care unit:

1.Oxygen administrated arbitrarily in concentrations that maintain SaO2 well above 95%.
2.Frequent airway suctioning via the tube.
3.Supplemental oxygen increased when desaturations occur.
4.Ventilator weaning attempted at the expense of hypercapnia.
5.Extubation not attempted unless the patient appears to be ventilator weaned.
6.Extubation to CPAP or low span bi-level positive airway pressure and continued oxygen therapy.
7.Deep airway suctioning by catheterizing the upper airway along with postural drainage and chest physical therapy.
8.With increasing CO2 retention or hypoxia supplemental oxygen is increased and ultimately the patient is reintubated.
9.Following re-intubation tracheostomy is thought to be the only long-term option...or following successful extubation bronchodilators and ongoing routine chest physical therapy are used.
10.Eventually discharged home with a tracheostomy, often following a rehabilitation stay for family training.

We have found the following protocol to be quite successful in PREVENTING TRACHEOSTOMY and MAINTAINING NON-INVASIVE ventilation:

1.Oxygen administration limited only to approach 95% SaO2.
2.Mechanical insufflation-exsufflation (Cough Assist) used via the tube at 25 to 40 cm H2O to -25 to -40 cm H2O pressures up to every 10 minutes as needed to reverse oxyhemoglobin desaturations due to airway mucus accumulation and when there is ascultatory evidence of secretion accumulation. Abdominal thrusts are applied during exsufflation. Tube and upper airway are suctioned following use of expiratory aids as needed.
3.Expiratory aids used when desaturations occur.
4.Ventilator weaning attempted without permitting hypercapnia.
5.Extubation attempted whether or not the patient is ventilator weaned when meeting the following:

- ♥ afebrile
- ♥ no supplemental oxygen requirement to maintain SaO2 >94%
- ♥ chest radiograph abnormalities cleared or clearing
- ♥ any respiratory depressants discontinued
- ♥ airway suctioning required less than 1-2x/eight hours
- ♥ coryza diminished sufficiently so that suctioning of the nasal orifices is required less than once every 6 hours (important to facilitate use of nasal prongs/mask for post-extubation nasal ventilation)

6.Extubation to continuous nasal ventilation and no supplemental oxygen.
7.Oximetry feedback used to guide the use of expiratory aids, postural drainage, and chest physical therapy to reverse any desaturations due to airway mucus accumulation.
8.With CO2 retention or ventilator synchronization difficulties nasal interface leaks were eliminated, pressure support and ventilator rate increased or the patient switched from BiPAP-STtm to using a volume cycled ventilator. Persistent oxyhemoglobin desaturation despite eucapnia and aggressive use of expiratory aids indicated impending respiratory distress and need to re-intubate.
9.Following re-intubation the protocol was used for a second trial of extubation to nasal ventilation...or following successful extubation bronchodilators and chest physical therapy were discontinued and the patient weaned to nocturnal nasal ventilation.
10.Discharge home after the SaO2 remained within normal limits for 2 days and when assisted coughing was needed less than 4 times per day.

When extubating, keep in mind that the throat is swollen. It may be needed to replace the tube with a smaller one, give steroids to diminish swollen, and then try extubation again. Follow with aggressive CPT, suction, and cough-a-lation every 4 hours or as needed.

BIBLIOGRAPHY

NIV INFORMATION

From Non-Invasive Mechanical Ventilation Center at University Hospital, NJ As presented by Dr. John Bach Jr. from UMDNJ

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John R. Bach, MD, Professor of Physical Medicine and Rehabilitation, Vice-Chairman of the Department of Physical Medicine and Rehabilitation, Professor of Neurosciences, Director of Research and Associate Medical Director of the Department of Physical Medicine and Rehabilitation, Co-Director of the MDA Clinic at University Hospital, and Medical Director of the Center for Noninvasive Mechanical Ventilation Alternatives and Pulmonary Rehabilitation.

Dr. Bach is **internationally** recognized for his ground-breaking work in noninvasive mechanical ventilation. He has been listed in several physician-selected consumer guides to outstanding physicians, including the "Best Doctors in America" by Castle, Connelly and New York Magazine's "Best Doctors in New York".

The author of over 250 publications, including seven books on neuromuscular pulmonary rehabilitation and noninvasive mechanical ventilation, Dr. Bach has been invited to lecture on these topics in over 30 countries.

Dr. Bach is a fellow of the Association of Academic Physiatrists, the America College of Chest Physicians, and the American Academy of Physical Medicine and Rehabilitation. He received his medical degree from the College of Medicine and Dentistry of New Jersey in 1976 and completed his residency training in Physical Medicine and Rehabilitation at New York University.

MORE SMA INFORMATION

Diagnosing SMA

Physical Characteristics:

The common physical characteristics of SMA include:

- "Frog" shaped legs (knees apart and legs bent)
- a sunken or narrow chest
- a big belly
- breathing with the belly instead of the lungs
- a weak cry and weak cough
- poor to no head/neck control; head tilted to one side
- weak if any movement of the legs and upper arms
- not able to bear any weight on legs or arms
- hands that remain clenched or turned the wrong way
- difficulty sucking and swallowing
- tongue fasciculations (tongue vibrating rapidly).

These characteristics vary in severity depending on how advanced the SMA is in each child. Some of these characteristics may not show up until later.

Medical Testing:

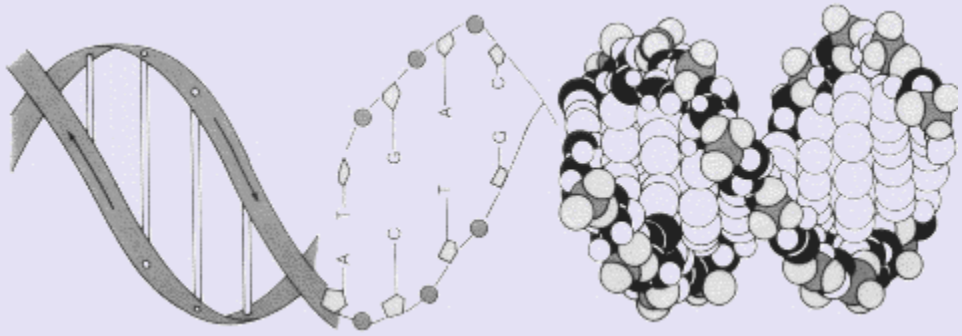
There are several medical tests that can be performed to diagnose SMA. Following are a listing of the tests along with an explanation of how they are performed and their accuracy:

- **EMG (Electromyography)**
  An EMG test measures the electrical activity of muscle. In this procedure small needles are gently inserted into the patient's muscles (usually the arms and thighs) while an electrical pattern is observed and recorded by a specialist.
- At the same time, a **nerve conduction velocity (NVC)** will probably also be performed. This uses the same needles and equipment. In this test the response of a nerve to an electrical stimulus is measured. When performing this test on a child, if at all possible, it should be performed by a doctor experienced in dealing with children. If permitted, hold your child on your lap during the procedure, to make an unpleasant procedure somewhat bearable. Your doctor may allow your child to be given a mild pain killer or sedative prior to the test.
- **Genetic Blood Test**
  Within the last decade, a blood test has now become available to detect SMA. This blood test works by detecting deletions in gene sequences that

are not missing in normal, healthy individuals. This blood test can not tell the Type of SMA that the individual has (Type I, II, or III), and approximately 5% of individuals who do have SMA do not show the gene deletions. However, for the 95% of individuals who do show the deletions, the diagnosis is 100% accurate, and the Type of SMA can be determined by other physical factors. With a blood test to screen for SMN deletion together with an EMG and a clinical examination it may not be necessary for a muscle biopsy to be performed. If the results show that there is no deletion of the SMN gene, but the clinical examination and the EMG still point to SMA, than a muscle biopsy would be necessary to confirm the diagnosis.

- **Muscle Biopsy**
  This is a surgical procedure where an incision approximately 3 inches long is made, and a small section of muscle is removed. Usually they remove the muscle from the upper thigh. The biopsy is used to check for degeneration of muscles and special tell-tale signs in the muscles of SMA. It is important to find a doctor used to dealing with children, and experienced in dealing with SMA. Although many doctors may persuade you of the necessity of a general anesthetic, this procedure can be done with a local anesthetic. This is especially important when dealing with children who are possibly suffering from SMA which includes by nature a weak respiratory system. General anesthesia is not recommended for children with neuromuscular diseases such as SMA as it may be difficult for them to recover.
- **Needle Biopsy**
  There is now an alternative to a muscle biopsy. Instead of a 2-3 inch incision, only a small nick in the skin is necessary. Be sure to ask your doctor about this possibility.

SMA in Scientific Terms



Spinal Muscular Atrophy (SMA) is one of the neuromuscular diseases. Muscles weaken and waste away (atrophy) due to degeneration of anterior horn cells or motor neurons which are nerve cells in the spinal cord. Normally, these motor neurons relay signals, which they receive from the brain, to the muscle cells. When these neurons fail to function, the muscles deteriorate. SMA effects the voluntary muscles for activities such as crawling, walking, head and neck control and

swallowing.

SMA mainly affects the proximal muscles, or in other words the muscles closest to the trunk of the body. Weakness in the legs is generally greater than weakness in the arms. Some abnormal movements of the tongue, called tongue fasciculation's may be present in patients with Type I and some patients with Type II. The brain and the sensory nerves (that allow us to feel sensations such as touch, temperature, pain etc.) are not affected. Intelligence is normal. In fact it is often observed that patients with SMA are unusually bright and sociable.

Type 1 Severe Infantile SMA, or Werdnig-Hoffman disease

Infantile spinal muscular atrophy (Werdnig-Hoffman disease) is the most severe form of SMA. It usually becomes evident in the first six months of life. The child is unable to roll or sit unsupported, and the severe muscle weakness eventually causes feeing and breathing problems. There is a general weakness in the intercostals and accessory respiratory muscles (the muscles situated between the ribs). The chest may appear concave due to the diaphragmatic breathing. These children usually do not live beyond about 24 months of age.

Type 2 Intermediate type

A child with the intermediate form of SMA often reaches six to twelve months of age, sometimes later, and learns to sit unsupported, before symptoms are noticed. Weakness of the muscles in the legs and trunk develops and this makes it difficult for the child to crawl properly or to walk normally, if at all. Weakness in the muscles of the arms occurs as well although this is not as severe as in the legs. Usually the muscles used in chewing and swallowing are not significantly affected early on. The muscles of the chest wall are affected, causing poor breathing function. Parents notice that the child is "floppy" or limp, the medical term for this being hypotonia. Tongue fasciculations are less often found in children with Type II but a fine tremor in the outstretched fingers is common. Children with Type II are also diaphragmatic breathers. Physical growth continues at a normal pace and, most importantly, mental functions is not affected. The children are bright and alert and it is important that they receive all the available opportunities to develop their intellectual capacities to their fullest extent. Integration into a normal school environment gives them the best chance to mature intellectually and emotionally.

What does the future hold?

The course of the disease is quite variable, and difficult to precisely predict from the start. Children with the intermediate form of SMA usually sit unsupported. Weakness of the legs and trunk usually, but not always, holds the child back from standing and walking alone. Sometimes the muscle weakness can seem to be non-progressive, but in most cases weakness and disability will increase over many years. Severe illness with prolonged periods of relative immobility, putting on excessive weight or growth spurts may contribute to deterioration in function. Due to weakness of the muscles supporting the bones of the spinal column, scoliosis

(curvature of the spine) often develops in children who are wheelchair bound. If this becomes severe it can cause discomfort and can have a bad influence on breathing function as well. An operation can be done to straighten the spine and prevent further deterioration. Recurrent chest infections may occur, because of decreased respiratory function and difficulty in coughing. Parents will have been shown how to encourage their child to maintain his/her maximum respiratory function as well as how they can perform postural drainage of the chest. They should start this as the first sign of any chest problem. Antibiotics and inhalation therapy may also be needed. Sometimes hospitalization is required to best manage and care for the child. The long term outlook depends mainly on the severity of weakness of the muscles of the chest wall and on the development of scoliosis. Lifespan is always difficult to predict. Mildly affected children may live into adult years. The more severely affected children may die, due to pneumonia and other chest problems, before or in their teens.

Type 3 Mild Juvenile SMA, or Kugelberg-Welander disease

Juvenile spinal muscular atrophy (Kugelberg-Welander disease) usually has its onset after 2 years of age. It is considerably milder than the infantile or intermediate forms. In juvenile spinal muscular atrophy children are able to walk, although with difficulty. The patient with Type III can stand alone and walk, but may show difficulty with walking and/or getting up from a sitting or bent over position. With Type III, a fine tremor can be seen in the outstretched fingers but tongue fasciculations are seldom seen.

Type 4 Adult Onset

Typically in the adult form symptoms begin after age 35. It is very rare for Spinal Muscular Atrophy to begin between the ages of 18 and 30. Adult SMA is characterized by insidious onset and very slow progression. The bulbar muscles, those muscles used for swallowing and respiratory function, are rarely affected in Type IV.

Type 5 Kennedy's Syndrome or Bulbo-Spinal Muscular Atrophy

This form also known as Adult Onset X-Linked SMA, occurs only in males, although 50% of female offspring are carriers. This form of SMA is associated with a mutation in the gene that codes for part of the androgen receptor and therefore these male patients have feminine characteristics, such as enlarged breasts. Also noticeably affected are the facial and tongue muscles. Like all forms of SMA the course of the disease is variable, but in general tends to be slowly progressive or non-progressive.

SMA in Laymen's Terms

SMA is a muscular disease passed on genetically to children by their parents. You can not "catch" SMA by being around someone who has it. It is a "Recessive"

genetic disease, meaning that BOTH parents must carry a copy of the recessive SMA gene. There is only a 25% chance each pregnancy of the child having SMA and a 75% chance each pregnancy that the child will be healthy. One out of 40 people is a carrier of this recessive gene. The brain is not affected, and they have been tested to have at least average to above average intelligence. Please do not make the mistake of treating them as mentally impaired!! Their bodies may not be perfect, but their minds are, so be sure to treat them that way! SMA affects a child's muscular development, and the severity depends on what 'type' of SMA the child has. There are four "Types" of SMA, Type 1,2,3 & 4. The earlier the symptoms are noticed, the more severe the type of SMA. Type 1 is the most severe, affecting children while still in the womb or shortly after birth. Type 4 is the least severe, affecting adults.

Type 1 children are diagnosed usually before 6 months of age, more often before 3 months of age. Symptoms may even start in the womb. Many mothers later recall the baby not moving as much the last month or so of pregnancy. They are not able to hold up their heads, roll over, crawl, sit up without support, or walk. All of their muscles are extremely weak, with the weakest muscles being the legs, upper arms, and neck. Their chest may appear concave, or very skinny at the top, with a big belly. Bell-shaped. SMA affects all muscle systems as well including sucking, swallowing, digesting food, and excretion. Constipation is a common problem as is being able to control excessive drooling (secretions), and getting proper nutrition and calories for proper weight gain. A common cold can easily turn into pneumonia which is what usually takes the lives of these children, along with "respiratory failure" or when they no longer have the lung or chest muscles to be able to breathe on their own. Two major decisions must be made with Type I children...whether or not to insert a feeding tube to prevent pneumonia and prevent starvation when they have lost their ability to suck or swallow; and whether or not to put them on a ventilator or other breathing machine when they experience respiratory failure. Current statistics show that the average lifespan of a child with SMA Type I, not put on permanent ventilation or "life support", is only 8 months of age, with 80% dying by the age of one, and the majority of the rest dying by age 2. HOWEVER, these statistics are not a hard and fast rule. Each child is affected so differently by SMA that they do not all follow the same path or progression. Also, as more is learned about SMA, the lifespan of a Type 1 child can be lengthened depending on the severity of the symptoms for each particular child. Last but not least, the line between each Type of SMA is not clearly defined, and it is common for a child to exhibit patterns of two types, thus confusing the issue of "life expectancy" for that child.

Type I children most often have very little leg movement, very little upper arm movement. Many times their hands remain fisted and their hands/wrists are turned the "wrong" way. The physical characteristics that often "gives them away" to having SMA is a bell shaped body, legs that stay in the "frog" position, moving the arms from the elbows down only, and the head tilted to the side because of lack of neck muscles. They often have bright, expressive faces and eyes.

Type 2 children are diagnosed before 2 years of age, usually more like 15 months. These children are usually able to be in a sitting position without support, but often can not get there by themselves. They can sometimes crawl with bracing and therapy, and on occasion may stand with braces. Feeding and swallowing problems are not common in Type 2 children, though they are still possible. They will usually never walk. The lifespan of a Type 2 child varies so widely, there isn't one! They could pass away at an early age or they could live well into adulthood. As with all forms of SMA, weakness increases over time.

Type 3 children are diagnosed between 18 months of age and early adolescence. In the beginning these children are able to stand and walk but usually have difficulty doing so. They typically have a normal lifespan; however, as with all forms of SMA, weakness gets progressively worse and they usually will be wheelchair bound.

Type 4 SMA is an adult SMA, with symptoms beginning around age 35. They also usually have a normal lifespan; though, as with all forms of SMA, weakness gets progressively worse.

SMARD: Info from JTSMA

Spinal Muscular Atrophy with Respiratory Distress Type 1 (SMARD1) is a neuromuscular condition causing weakness of the muscles. It is the second anterior horn cell disease in infants in which the genetic defect has been defined. SMARD1 is not linked to the SMN1 gene locus on chromosome 5q13 (classic SMA), but is caused by mutations in the IGHMBP2 gene on chromosome 11q13. Despite a substantial overlap in clinical features, the phenotypes of SMA versus SMARD1 can be distinguished. In SMARD1, the predominating symptom is a severe respiratory distress due to a paralysis of the diaphragm. Most patients present [show symptoms] at the age of 1 to 6 months with respiratory failure and progressive muscle weakness with predominantly distal lower limb muscle involvement. Sensory and autonomic nervous systems may also be involved. Early involvement of the diaphragm and predominance of distal muscle weakness clearly distinguishes SMARD1 from Type 1 SMA. Essentially, in SMA1, symptoms manifest in reverse order. Infants with SMA1 will become floppy due to weakness of the proximal limb muscles and assume a frog leg position before they suffer from respiratory failure. In contrast to SMARD1, SMA1 infants have intercostal recessions and develop inefficient respiration due to paralysis of intercostal muscles.

Parents dealing with a new diagnosis of SMA Type 1 in their children have three basic options of care/treatment.

1.Palliative Care- This is the option parents choose if they do not wish to intervene in their child's illness. A child with SMA Type 1, with no intervention, has a life expectancy of approximately 8 months, with 80% gone by 1 year. Palliative Care is also known as "Comfort Care". Parents will choose to use oxygen and pain medications such as morphine to help keep the child as comfortable as possible

until they pass away. This method is for parents who believe their child's quality of life would be poor, so they choose out of love to let nature take its course and enjoy their children with the time they are given.

2.NIV Protocol- Also known as **"Dr. Bach's Non-Invasive Protocol"**. This is a respiratory protocol using a breathing machine called "Bipap" and a machine called a "Cough Assist" to help manage the respiratory issues that are core problems with SMA. Parents will choose to use these machines as a method of providing respiratory comfort and strength for their child. The life expectancy of a Type 1 SMA child using the NIV protocol is currently unknown and completely depends on other circumstances as well. It is not a cure for SMA, there are no guarantees in life or SMA, and the children will still be profoundly handicapped, but it has shown to increase the quantity and quality of many children's lives to date. The oldest known true Type 1's surviving using these methods were born in 1996. This method is chosen out of love by parents who do not at the time wish to pursue a surgical option but do not wish to let their child go, and believe they can provide a good quality of life for their child and believe that what they are doing improves the quality and comfort of their child's life, also attempting to prolong their child's life out of hope for the future.

3.Tracheotomy- This is a surgical procedure, creating a hole in the throat that provides breathing assistance via a permanent ventilator. The life expectancy of an SMA Type 1 child with a trach is unknown and completely depends on other circumstances as well. It is not a cure for SMA, there are no guarantees in life or SMA, and the children will still be profoundly handicapped, but it is possible to survive to adulthood. This method is for parents who believe they can provide a good quality of life for their child now and in the future, so they choose out of love to prolong their child's life indefinitely out of the belief that their child is happy and simply receiving respiratory support, and out of hope for the future.

Werdnig-Hoffman disease / Spinal Muscular Atrophy Type 1

Definition from Wikipedia

Werdnig-Hoffman disease (also known as "Infantile spinal muscular atrophy", "spinal muscular atrophy type 1", or "spinal muscular atrophy type I") is an autosomal recessive muscular disease. It is the most severe form of spinal muscular atrophy. Werdnig-Hoffman affects the lower motor neurons only. It has been linked to an abnormal survival motor neuron (SMN) gene.

Symptoms

It is evident before birth or within the first few months of life. There may be a reduction in fetal movement in the final months of pregnancy. Symptoms include floppiness of the limbs and trunk, feeble movements of the arms and legs, swallowing and feeding difficulties, and impaired breathing. Affected children never

sit or stand unassisted and will require respiratory support to survive before the age of 2. Other symptoms include:
•Fasciculations of the tongue
•Marked Hypotonia in Proximal, Distal muscles, Intercostals & bulbar muscles (Patient lies in a Frog-Leg position,i.e. hips abducted & knee flexed)
•Flaccid Quadriplegia
•Difficulty breathing
•Poor feeding - need gastrostomy
•Weak cry
•Areflexive extremities
•Normal Intelligence

Diagnosis
•Electro-Myelo Gram (EMG) will show Fibrillation & Muscle Denervation
•Serum Createnine-Kinase may be normal or Increased

Treatment
Treatment is symptomatic and supportive and includes treating pneumonia, curvature of the spine and respiratory infections, if present. Also, physical therapy, orthotic supports, and rehabilitation are useful. For individuals who survive early childhood, assistive technology can be vital to providing access to work and entertainment. Genetic counseling is imperative.

Prognosis
The patient's condition tends to deteriorate over time, depending on the severity of the symptoms. Children with Werdnig-Hoffman disease / SMA Type 1 face a difficult battle. They are constantly at risk of respiratory infection and pneumonia. Feeding difficulties make it a real challenge for parents to give their children adequate nutrition and supplemental feedings may be required. Tubes placed through the nose or directly onto the stomach may be necessary. Recurrent respiratory problems mean that mechanical support for breathing - usually initially in the form of BiPAP and later often tracheostomy and ventilation - are necessary for the baby to have any chance of long-term survival. Affected children never sit or stand and usually die before the age of 2 if the decision is made not to provide breathing support. However, some individuals have survived to become adults, in which case sexual function is unimpaired.

Nutrition: A New Understanding

Muscle Metabolism in Spinal Muscular Atrophy and Other Muscle Disorders

Atrophy or severe underdevelopment of muscle is common to SMA and many other neuromuscular diseases. Although the most obvious result of muscle atrophy is weakness, reduced muscle mass also has serious effects of nutrition, because muscle functions as an essential nutritional reserve, or "buffer," for protein, carbohydrate, and mineral metabolism. The loss or reduction of this buffering capacity limits the ability of the body to adjust to simple nutritional changes, such as normal overnight fasting, or to more serious threats, such as diarrhea, vomiting, and fevers. For example, the limited ability of atrophied muscle to replace losses of potassium and water is one of the main reasons that an otherwise simple diarrhea illness can be lethal to a malnourished child or adult.

The role of muscle in the metabolism of fasting is especially important. Although the liver has a reserve of glucose in the form of "glycogen" that can sustain blood sugar levels for six to eight hours after a meal, muscle becomes the primary source of glucose for longer periods of fasting. Muscle does this by degrading its own protein into amino acids and then releasing the amino acids to the bloodstream. There the amino acids are picked up by the liver and turned into glucose. The glucose that is made in this manner is vital for brain and nerve function, and the body will sacrifice as much muscle protein as necessary to maintain blood glucose levels during fasting.

For the average child or adult, the amount of muscle protein that is degraded in one day of fasting may be only one percent of the total muscle mass. However, for a child with SMA who may have only 10% of normal muscle mass, a much greater proportion of muscle mass must be sacrificed to supply amino acids for glucose synthesis. Even the small net loss of protein that occurs during normal overnight fasting may be a significant loss for a child with SMA who not only has limited muscle mass during the daytime. Blood sugar and amino acid levels are the major signals for muscle protein metabolism: if they are high there is net muscle protein synthesis, and if they are low, as during fasting, there is net muscle protein breakdown. We have studied the adaptation to fasting of children with SMA for several years to learn more about muscle metabolism and nutrition in SMA.

We first confirmed that within only two or three hours of a normal meal or feeding, the blood amino acid levels of children with all forms of SMA decreased to levels that would not be reached until after at least eight hours of fasting in a normal child. We were also surprised to find that children with infantile or "acute" SMA could not efficiently metabolize fatty acids, which are a major source of energy during periods of fasting. Rather, children with acute SMA had high levels of unused (and possibly toxic) fatty acid by-products in their urine and blood, sometimes after only overnight fasting. This abnormal fatty acid metabolism persisted in some

children with SMA who were hospitalized for an illness and receiving glucose intravenously. At this time we cannot explain all of the biochemical abnormalities that we see in SMA, nor can we prescribe a precise remedy for the problem. Nevertheless, following basic principles of normal muscle nutrition and metabolism may help preserve muscle mass in children with SMA.

The goals of such nutritional therapy should be to 1) limit fasting and the resulting low levels of blood glucose and amino acids that turn on muscle protein breakdown, and 2) assure adequate amounts of dietary protein to maintain normal blood amino acid levels and thereby enhance protein synthesis by muscle.

A suitable nutritional program to achieve these two simple goals would include 1) increased amounts of complex carbohydrates, 2) at least 2 grams of protein per kg per day, and 3) a feeding schedule that limits overnight fasting to 6 hours for a young infant and 10-12 hours for an older child. Of course, full caloric requirements for each child's age and level of activity must also be met. Such a diet will necessarily have a lower percentage of fat, but is also important to assure that a child receives an adequate amount of essential fatty acids (a minimum of 5% of calories). For older infants and children with SMA who sleep through the night, a late evening supplement of uncooked cornstarch (1 gm/kg) with a bottle or other food provides a good source of slowly absorbed complex carbohydrate. Uncooked cornstarch sustains blood glucose levels longer than other starches and, in effect, reduces the period of overnight fasting.

Finally, careful attention to nutrition must be given during illnesses, especially those that cause vomiting or otherwise limit caloric intake. If a child with SMA cannot eat or drink to supply the daily requirement of calories (typically 100Kcal/kg/day for an infant, 70-80 kcal/kg/day for an older child) then hospitalization for high caloric intravenous glucose (10% dextrose at 1.5 times "maintenance") may be necessary.

Although we recognize that nutritional "therapy" is not a definitive treatment for SMA or any other muscle disease, following these principles of muscle nutrition may help limit unnecessary losses of muscle mass. This approach might help maintain strength longer and enhance the important buffering function of muscle for many different aspects of body nutrition and metabolism.

We hope to explore these areas of muscle metabolism in SMA in more detail in the future. Perhaps then we and others who study SMA may be able to provide more detailed guidelines for nutrition for children with SMA.

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Reprinted from Direction, Fall 1993, Vol. 7, Issue 4.

WHAT IS THE AMINO ACID DIET?

ABOUT THE DIET

The Amino Acid Diet (AAD) is a nutritional approach that many SMA families believe has had a positive impact on their child's wellness and stability. Using this dietary approach, milk, soy, and most other food proteins are eliminated and substituted with an elemental free form amino acid formula. Elemental formulas are made from nutrients in their most broken down form. This form requires very little digestion. Tolerex and Pediatric Vivonex, manufactured by Novartis Nutrition, are the two formulas that are used most often. Both formulas contain vitamins, minerals, carbohydrates, protein and a small amount of fat. The protein is in the form of free amino acids. A free amino acid is not derived from a food source and is in its simplest and purest form. Parents that use this diet believe more benefit is seen with free form amino acids, rather than hydrolyzed amino acids, which are broken down proteins from foods, such as dairy or soy. The free form amino acids in Pediatric Vivonex and Tolerex are in balanced amounts, and will not trigger immune responses such as inflammation. They are easily digested and quickly absorbed into the bloodstream. Many parents believe their children with SMA are extraordinarily reactive to the proteins in dairy and soy products. Their symptoms are commonly attributed to their SMA progression, yet when these proteins are eliminated and substituted with free form amino acids, most children, with all types of SMA, have experienced improved respiratory health and fewer complications from illness. Most children with Type 1 SMA have an immediate reduction in airway secretions. In addition, constipation, which can be a major complication, is more manageable or even eliminated. Some children have improved strength and regained function.

Parents using these formulas may vary their child's particular diet based on age, weight and severity of SMA, but there are some basics that most children respond to favorably. Although these formulas can be used to provide complete nutrition, many SMA families use approximately 1/3-1/2 of this amount in order to avoid amino acid overload. Children with SMA do not tolerate large amounts of amino acids due to their reduced muscle mass. This reduction of muscle mass prohibits the proper absorption and storage of amino acids resulting in possible toxic levels in the bloodstream. More than 2 packets of either formula, over a 24 hour period, can cause an increase in heart rate, other cardiac problems and/or gastrointestinal distress. To avoid protein and calorie overload, these formulas should not be given to a child still on soy or milk based formulas. The idea is to replace those food proteins with the free form amino acid protein found in the Tolerex or Pediatric Vivonex.

ADDITIONAL SUPPLEMENTS

Because these formulas do not provide complete nutrition, additional vitamins and minerals must be added to the child's diet to ensure that the RDA's are met. Special

attention should be given to calcium, magnesium, and potassium intake. Other commonly well-tolerated added supplements (by most children): vitamins and minerals, acidophilus (dairy free), Lglutamine powder (dosage is 200-300mg/kg/day), (the formulas contain 1060mg's in one pack of Tolerex and 774mg's in one pack of Pediatric Vivonex), L-Carnitine, and Coenzyme Q10. Sometimes, small amounts of additional essential amino acids, in free form, are used by older children.

Although children with SMA do not require as many calories as active children without SMA, the amount of formula used is inadequate to meet their caloric need. If using Tolerex, parents sometimes add very small amounts of extra fat to ensure that adequate essential fats are given. A commonly used fat supplement is 1/4 to 1 teaspoon of safflower oil added to the daily formula mixture. Some children do well with 1-2 grams of evening primrose oil per day. Any fat supplements should be added gradually over a period of days, as some of the most affected children may experience slow stomach emptying and gastric distress. The Pediatric Vivonex contains ample fat for most children.

INSTRUCTIONS ON ADMINISTERING DIET

The Formula:
These formulas should be diluted as much as possible (more than the directions call for) with water, juices or enriched rice milk. The benefits of using juices or rice milk include not just the improved palatability, but the additional calories, potassium, and other nutrients they provide. A well-tolerated dilution is approximately 12-20 ounces of liquid for each packet of formula, depending on whether the child is G-tube fed or drinking the formula. To ensure adequate hydration, extra fluids should be given. Children that are exclusively G-tube fed should have the formula administered by a continuous drip or given in small boluses. Some children without G-tubes drink the formula throughout the day. It should be diluted as much as possible, and divided into at least 3-4 servings.

Additional Food:
In addition to their Tolerex or Pediatric Vivonex, SMA children should consume fruits, vegetables, and those with types 2 or 3 SMA usually tolerate grains. Parents should use vitamins, minerals, additional supplements (discussed earlier), juices, and baby foods (fruit and vegetable) to meet their child's nutritional requirements. The formula, fluids, and any added supplements and baby foods should be mixed in a blender and refrigerated. . Since children with Type 1 SMA are often the most reactive to changes in their diet, foods and supplements should be added one at a time to check for tolerance. Children receive the most benefit from the diet by greatly reducing or avoiding animal protein foods. This includes dairy products, eggs, and meat. A vegetarian diet is highlyrecommended to see the most benefit. Remember, the children are receiving adequate protein from the amino acids in their elemental formula, and can easily be overloaded if they are also eating high-protein foods. For some children, this can interfere with improvements they may gain from this diet.

CLOSING

Personally, I have had success with this diet for my daughter Krista, a happy, vibrant 15-year old who has SMA Type 2. Through research, trial-and-error, we placed Krista on this diet over 10 years ago and have been very pleased with her wellness and stability. Although children are affected by their SMA differently, the recommendations stated earlier seem to be the safest and most effective way to implement this diet. Parents must take responsibility for monitoring and problem solving when placing their child on this diet. Unfortunately, due to the lack of study this diet has received, most medical professionals are unfamiliar with this dietary approach and often overload our children with these formulas. I encourage all parents to research and read the many informative nutrition books available at bookstores and libraries. In addition, networking with other SMA families that have successfully used this diet can also be an invaluable source for information on the benefits and administration of this regimen.

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This information is based on opinions and experiences compiled from parents of children with SMA. This diet has not been studied by the scientific community and is not endorsed by Families of SMA.

ADDITIONAL INFORMATION

Pediatric Vivonex and Tolerex are formulas manufactured by Novartis Nutrition. Ph# 1-800-333-3785. With a prescription, they are often paid for by insurance or Medicaid. Information *on their home delivery program can be found at* *www.resource.walgreens.com*

Aminoacid based diets:
ELECARE= 33grams of fat
NEOCATE= 23 grams of fat
NEOCATE+1= 14 grams of fat
VIVONEX= 12 grams of fat in a 1800 complete feeding
TOLEREX= 2.6 grams of fat in a 1800 complete feeding

APENDIX 2

CONSENSUS STATEMENT for STANDARD of CARE in SPINAL MUSCULAR ATROPHY AUGUST 2007 (MDA QUEST NOV 2007)

FAMILY GUIDE to the CONSENSUS STATEMENT

UW Health Facts: COUGH ASSIST MACHINE

COUGH ASSIST STUDIES

CHOICES PRESENTATION

Journal of Child Neurology
http://jcn.sagepub.com

Consensus Statement for Standard of Care in Spinal Muscular Atrophy

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J Child Neurol 2007; 22; 1027
DOI: 10.1177/0883073807305788

The online version of this article can be found at:
http://jcn.sagepub.com/cgi/content/abstract/22/8/1027

Published by:
SAGE Publications
http://www.sagepublications.com

Special Issue Article

Journal of Child Neurology
Volume 22 Number 8
August 2007 1027-1049
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10.1177/0883073807305788
http://jcn.sagepub.com
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http://online.sagepub.com

Consensus Statement for Standard of Care in Spinal Muscular Atrophy

Ching H. Wang, MD, PhD, Richard S. Finkel, MD, Enrico S. Bertini, MD, Mary Schroth, MD, Anita Simonds, MD, Brenda Wong, MD, Annie Aloysius, MRCSLT, HPC, Leslie Morrison, MD, Marion Main, MCSP, MA, Thomas O. Crawford, MD, Anthony Trela, BS, and Participants of the International Conference on SMA Standard of Care

Spinal muscular atrophy is a neurodegenerative disease that requires multidisciplinary medical care. Recent progress in the understanding of molecular pathogenesis of spinal muscular atrophy and advances in medical technology have not been matched by similar developments in the care for spinal muscular atrophy patients. Variations in medical practice coupled with differences in family resources and values have resulted in variable clinical outcomes that are likely to compromise valid measure of treatment effects during clinical trials. The International Standard of Care Committee for Spinal Muscular Atrophy was formed in 2005, with a goal of establishing practice guidelines for clinical care of these patients. The 12 core committee members worked with more than 60 spinal muscular atrophy experts in the field through conference calls, e-mail communications, a Delphi survey, and 2 in-person meetings to achieve consensus on 5 care areas: diagnostic/new interventions, pulmonary, gastrointestinal/nutrition, orthopedics/rehabilitation, and palliative care. Consensus was achieved on several topics related to common medical problems in spinal muscular atrophy, diagnostic strategies, recommendations for assessment and monitoring, and therapeutic interventions in each care area. A consensus statement was drafted to address the 5 care areas according to 3 functional levels of the patients: nonsitter, sitter, and walker. The committee also identified several medical practices lacking consensus and warranting further investigation. It is the authors' intention that this document be used as a guideline, not as a practice standard for their care. A practice standard for spinal muscular atrophy is urgently needed to help with the multidisciplinary care of these patients.

Keywords: spinal muscular atrophy; standard of care; consensus statement

Current Problems in the Medical Care of Patients With Spinal Muscular Atrophy

Spinal muscular atrophy is a recessively inherited neuromuscular disease characterized by degeneration of spinal cord motor neurons, resulting in progressive muscular atrophy and weakness. The clinical spectrum of spinal muscular atrophy ranges from early infant death to normal adult life with only mild weakness. These patients often require comprehensive medical care involving multiple disciplines. There is, however, no published practice standard for the care of these patients. Disparity in family resources, medical practitioners' knowledge, and regional and cultural standards produces wide variation in care and clinical outcome. Spinal muscular atrophy, as a field, has recently seen major advances in molecular diagnosis and clinical therapeutics that have not been matched by wide understanding and application. Parents of children newly diagnosed with spinal muscular atrophy often seek care over the Internet or outside of their geographic area to obtain expert care needed for their children, albeit in a costly and inefficient manner. This also undermines trust in local practitioners and their potential to render good clinical care during acute illness. Another consequence of these variations in practice is loss of trust in traditional medicine and increase in the attractiveness of untested and potentially harmful unconventional therapies. The large variation of clinical care in spinal muscular atrophy also

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results in challenges for future therapeutic trials. For all these reasons, we have identified an urgent need to establish a practice guideline, both to improve patient care and to provide a baseline standard for future clinical trials in spinal muscular atrophy.

The International Standard of Care Committee for Spinal Muscular Atrophy

Committee Formation

In September 2004, the National Institute of Neurological Diseases and Stroke sponsored an International Spinal Muscular Atrophy Conference in Bethesda, Maryland, with the goal of formulating strategies to coordinate future clinical trials in spinal muscular atrophy.[1] During the conference, it became clear that the wide variation of medical care received by spinal muscular atrophy patients likely increases the variability of outcomes in clinical trials. Thus, this Spinal Muscular Atrophy Standard of Care Committee was formed in January 2005, as a standing committee of the International Coordinating Committee for Spinal Muscular Atrophy clinical trials, to investigate the current state of science in clinical practice in spinal muscular atrophy and to attempt to achieve consensus on the standard of care for these patients.

Committee Structure

The Standard of Care Committee for Spinal Muscular Atrophy is cochaired by a US and a European neurologist. There are 12 core members and 3 consultants on the committee. Eleven of the core members are currently practicing pediatric neurologists, and 1 is a pediatric pulmonologist. The 3 consultants consist of 1 National Institute of Neurological Diseases and Stroke liaison and 2 representatives from patient advocacy groups. The committee is subdivided into 4 working groups: diagnostics/new interventions, pulmonary, gastrointestinal/nutrition, and orthopedics/rehabilitation. Each group is headed by a leader from the United States and a coleader from Europe. All committee members participated voluntarily, without pay. They were either nominated by their peers or have volunteered themselves for this task force. Each working group is composed of 6 to 11 experts in the field for that particular care issue (please see the Web site http://smascc.stanford.edu for a current roster of committee members).

Committee Missions

The committee has identified the following goals for all 4 working groups: (1) to identify current care issues in spinal muscular atrophy clinical practice, (2) to search for existing practices in spinal muscular atrophy clinical care and the rationale or data supporting such practices, (3) to achieve consensus of the most appropriate medical practice in caring for patients with spinal muscular atrophy, (4) to use this standard of care consensus to establish clinical care guidelines for future spinal muscular atrophy clinical trials, (5) to identify future research directions in the care of patients with spinal muscular atrophy, and (6) to publish the consensus as guidelines for clinical care of patients with spinal muscular atrophy.

Methods of Achieving Consensus on Standard of Care for Spinal Muscular Atrophy

Standards of medical practice are ideally established upon evidence-based clinical trial data. Unfortunately, committee core members found little data from well-designed clinical studies upon which evidence-based practice parameters in spinal muscular atrophy could be drafted (please see literature reviews in each care topic in the following sections). The absence of well-designed clinical trial data requires substitution of widely held opinion drawn from a survey of experts in the field. It is hoped that this consensus statement will serve both as an initial practice guideline for the care of spinal muscular atrophy and an outline of areas where needed clinical investigation may be best focused. We describe here the process leading to the drafting of this consensus statement.

Periodic Conference Calls and Literature Review

Since the inception of the committee in early 2005, the members have held periodic conference calls to discuss the ways to establish practice guidelines for spinal muscular atrophy. Group leaders were tasked with conducting literature reviews in their particular care areas. A password-protected Web site was established during this time. References of literature reviews were uploaded to the Web site. Having concluded that there were not enough published data to allow drafting of an evidence-based practice parameter, the group explored the possibility of using a Delphi survey to achieve consensus among experts in the field.

The Delphi Survey

The Delphi technique[2] was initially used to explore consensus expert opinion in government and education. More recently, it has been used in medicine, notably in rheumatology and neurology.[3-5] The goal of the Delphi technique is to identify if in aggregation there is a rank-ordered cluster of answers from respondents that reflects group consensus on that particular question. It also serves to identify if no consensus is present and where topics need further study. It presents group opinions anonymously, avoids domination by a few strong voices in the group, and can be completed by

electronic communications within a few weeks. Exploratory use of the Delphi technique was performed during an initial committee meeting in Philadelphia in June 2005. This served to familiarize the attending committee members with the mechanics of the Delphi technique and of its strengths and limitations. Having completed 2 rounds of pilot surveys among the committee members, the group concluded that the Delphi technique was suitable for establishing a consensus opinion among experts in spinal muscular atrophy. Group leaders then met by conference calls and e-mail communications to construct a formal Delphi survey questionnaire. During the first round of the Delphi survey, a set of open-ended questions was constructed for each of the 5 spinal muscular atrophy care topics (diagnostic/new interventions, pulmonary, gastrointestinal/nutrition, orthopedics/ rehabilitation, and palliative care). Each topic is divided into 3 parts: presenting signs and symptoms, diagnostic testing, and intervention options. The intervention part is then divided into acute management and health maintenance. These open-ended questions are named Question #1 (Q#1, available on the Web site http://smascc.stanford.edu). The Q#1 was distributed by ane-mail attachment to survey participants. A total of 86 spinal muscular atrophy experts were invited to participate in the survey. They were invited from 4 medical disciplines: 18 from the gastrointestinal/nutrition group, 21 from the pulmonary group, 25 neurologists from the diagnostic/new interventions group, and 22 from the orthopedics/rehabilitation group. Thirty-four of them were from Europe, and 52 were from the United States and Canada. All invited participants were recommended by committee members. The participants were allowed 3 weeks to respond to the questionnaire. Neurologists were encouraged to answer all 5 care topics. Respondents in the other 3 working groups generally limited their responses to respective areas of expertise. Fifty-six of the original 86 invited participants (65%) completed this Q#1. Twenty-two of them were from Europe, and 34 were from the United States and Canada. To ensure the anonymity of the process, a numeric code was assigned to each respondent by the survey coordinator upon receipt of answers to Q#1. Analysis and presentation of the data were performed by the survey coordinator using these numeric codes. The answers from Q#1 were collected and analyzed. The most frequent occurring answers to these Q#1 questions were chosen to construct the Question #2 (Q#2, available on the Web site http://smascc.stanford .edu) during the second round of the Delphi survey. In this second round, the questions were the same as those in Q#1 except that respondents were asked to rank order from the highest to the lowest importance among a list of choices. Forty-four (79%) respondents who answered the Q#1 also completed Q#2. These responses were summarized and presented to committee participants at the Standard of Care Conference described in the following section.

The International Conference on the Standard of Care for Spinal Muscular Atrophy

This conference was held May 5-6, 2006, at Stanford University Medical Center, Palo Alto, California. Thirty-five members of the committee and Delphi survey participants gathered to work on a consensus statement for spinal muscular atrophy standard of care. First, leaders and designated members of each working group presented a critical review of the literature. The individual working group then reviewed the results of the Delphi survey in their care areas during breakout sessions. The final consensus within each working group was achieved by using the Delphi data as a guideline, incorporating the available data in the literature and the opinions of group members. These results were presented by group leaders to all conference participants for comments. The group leaders and coleaders then worked with each working group to draft the consensus statement on each care area. The summaries of these statements are listed in the following sections.

Diagnostic Testing and Care of New Spinal Muscular Atrophy Patients

Clinical Diagnosis and Classification of Spinal Muscular Atrophy

Physicians encountering children with hypotonia and weakness should maintain a high index of suspicion for the diagnosis of spinal muscular atrophy. Certain physical characteristics are readily identifiable. The weakness is usually symmetrical and more proximal than distal. Sensation is preserved. Tendon reflexes are absent or diminished. Weakness in the legs is greater than in the arms. The severity of weakness generally correlates with the age of onset. The most severe type presents in infancy. The infant may appear normal at birth. Weakness evolves within the first few months of life. Occasionally, decreased intrauterine movements suggest prenatal onset of the disease and present with severe weakness and joint contractures at birth.[6] Milder types of spinal muscular atrophy present with later onset, and the course is more insidious. Some children sit but never walk, whereas others show delayed walking but may be able to maintain walking until adult years. For the purpose of clinical care and discussion, individuals manifesting different levels of weakness due to spinal muscular atrophy have been divided into 4 groups defined by functional ability. We list typical clinical features of spinal muscular atrophy in Table 1. The first 3 types are classified according to criteria established by the International Spinal Muscular Atrophy Consortium.[7,8] Type 4 spinal muscular atrophy is a mild form that presents in adulthood. It can be expected that some patients will manifest features that are at the margins between groups.

Table 1. Clinical Classification of Spinal Muscular Atrophy

SMA Type	Age of Onset	Highest Function	Natural Age of Death
Type 1 (severe)	0-6 mo	Never sits	<2 y
Type 2 (intermediate)	7-18 mo	Never stands	>2 y
Type 3 (mild)	>18 mo	Stands and walks	Adult
Type 4 (adult)	Second or third decade	Walks during adult years	Adult

NOTE: SMA = spinal muscular atrophy.

In addition to these defining criteria, unique clinical features of each spinal muscular atrophy type include the following: (1) Type 1 spinal muscular atrophy. This type is also called Werdnig-Hoffmann disease. Children with this disease have impaired head control, with a weak cry and cough. Swallowing, feeding, and handling of oral secretion are affected before 1 year of age. The tongue may show atrophy and fasciculation. Weakness and hypotonia in the limbs and trunks are eventually accompanied by intercostal muscle weakness. Combining intercostal weakness with initial sparing of the diaphragm, the infants exhibit characteristic paradoxical breathing and a bell-shaped trunk with chest wall collapse and abdominal protrusion. Early morbidity and mortality are most commonly associated with bulbar dysfunction and pulmonary complications. (2) Type 2 spinal muscular atrophy. These children have delayed motor milestones. Some learned to achieve independent sitting, whereas others need help to sit up. The defining characteristic is an ability to maintain a sitting position unsupported. At the strongest end of this category are those who can stand with a standing frame or long leg braces but are not able to walk independently. Bulbar weakness with swallowing difficulties may lead to poor weight gain in some children. Intercostal muscles are weak, and some are also diaphragmatic breathers. They have difficulty coughing and clearing tracheal secretion. They have fine tremors with extended fingers or when attempting hand grips. Kyphoscoliosis eventually develops, and bracing or spinal surgery is needed. Joint contractures commonly evolve over years. (3) Type 3 spinal muscular atrophy. This type is also called Kugelberg-Welander disease or juvenile spinal muscular atrophy. These patients have later but variable age of onset. All achieve independent walking. Some patients lose the ability to walk in childhood, yet others maintain walking until adolescence or adulthood. Scoliosis can develop in these patients. Swallowing, cough, and nocturnal hypoventilation are less common than in type 2 spinal muscular atrophy but may occur. Muscle aching and joint overuse symptoms are common. (4) Type 4 spinal muscular atrophy. The onset of weakness is usually in the second or third decade of life. Motor impairment is mild without respiratory or gastrointestinal problems.

Within each spinal muscular atrophy type, subclassifications have been proposed and can add to prognostic significance. For example, only 22% of patients with type 3a, with onset of symptoms before age 3 years, were still ambulatory at age 40 years, whereas 58.7% of the patients with type 3b, with onset after age 3 years, were still walking by age 40 years.[9] Type 1 patients have also been subclassified into types 1a (neonatal or antenatal onset), 1b (typical Werdnig-Hoffmann disease with onset after neonatal period), and 1c (later onset, better head control in supported sitting, mild feeding or respiratory difficulties during the first 6 months of life).[10,11] However, these subclassifications have not been widely used among clinicians.

During the preparation of the Delphi survey, the committee decided that the most appropriate care for patients with spinal muscular atrophy should be tailored according to their current functional status rather than the original classification of disease types because these represent the best level of function rather than the present status. Therefore, the committee decided to use the classification of current functional level in the form of nonsitters, sitters, and walkers. The nonsitters include the group of children who currently are not able to sit independently. The sitters include those who can sit independently but cannot walk independently. The walkers can walk independently.

Other Forms of Spinal Muscular Atrophy

There are other inherited motor neuron disorders, not caused by mutation of the *SMN* gene (non-5q spinal muscular atrophy), that present with early denervation weakness but different clinical symptoms than those stated above.[12] These atypical symptoms include joint contractures, distal rather than proximal weakness, diaphragmatic paralysis with early respiratory failure, and pontocerebellar degeneration. DNA testing has become available for some but not all of these disorders. If a child with clinical features of spinal muscular atrophy is found not to have an *SMN* deletion on either chromosome 5, the child should be reexamined and receive additional diagnostic testing. (Please see the next section for the diagnostic strategies for these patients.) Table 2 lists some spinal muscular atrophy variants that exhibit early symptoms overlapping with 5q spinal muscular atrophy. Several later-onset motor neuron diseases overlap with milder 5q spinal muscular atrophy. These are beyond the scope of this document and are not listed here.

Table 2. Other Forms of Severe Spinal Muscular Atrophy Not Linked to *SMN* Gene

SMA Variants	Inheritance/Linkage/Gene	Clinical Presentation	Reference
Scapuloperoneal spinal muscular atrophy	Autosomal dominant 12q24.1-q24.31	Congenital absence of muscles, progressive weakness of scapuloperoneal and laryngeal muscles	(13, 14)
Pontocerebellar hypoplasia with spinal muscular atrophy	Autosomal recessive	Onset 0-6 mo, cerebellar and brainstem hypoplasia, absent dentate nucleus, neuronal loss in basal ganglia, cortical atrophy	(15-19)
X-linked infantile spinal muscular atrophy with arthrogryposis	X-linked Xp11.3-q11.2	Onset at birth or infancy, contractures, death less than 2 y	(20, 21)
Spinal muscular atrophy with respiratory distress type 1	Autosomal recessive 11q13.2-q13.4 IGHMBP2	Onset within the first 3 mo of life, eventration of the right or both hemidiaphragms, finger contractures, pes equines foot deformities	(22, 23)

NOTE: SMA = spinal muscular atrophy.

Diagnostic Procedures

The stepwise algorithm of the diagnostic procedure is summarized in Figure 1. Briefly, the first diagnostic test for a patient suspected to have spinal muscular atrophy should be the *SMN* gene deletion test. This test is currently performed by several diagnostic laboratories, and the result can be obtained within 2 to 4 weeks. The test achieves up to 95% sensitivity and nearly 100% specificity.[24,25] A homozygous deletion of *SMN1* exon 7 (with or without deletion of exon 8) confirms the diagnosis of *SMN*-associated spinal muscular atrophy (5q spinal muscular atrophy). The next group of tests following a negative *SMN* test result includes repeat clinical examination of the patient for atypical clinical features as listed in Table 2. Laboratory tests should include muscle enzyme creatine kinase, electrophysiological testing such as electromyography (EMG), and nerve conduction study with repetitive stimulation. This will help to identify muscle diseases, motor neuropathies, and disorders of neuromuscular junctions. If EMG suggests a motor neuron disease, then further testing for *SMN* mutations should be pursued. Some laboratories are currently offering *SMN1* gene copy number testing. If the patient possesses only a single copy of *SMN1* (missing 1 copy), then it is possible that the remaining copy contains subtle mutations, including point mutations, insertions, and deletions, rendering homozygous dysfunction of the gene. Sequencing of the coding region of the remaining *SMN1* copy may identify the mutation on the remaining copy and confirm the diagnosis of 5q spinal muscular atrophy. Unfortunately, sequencing the coding region of *SMN* is currently not widely available and is usually performed only in a few diagnostic or research laboratories. If the patient possesses 2 copies of *SMN1*, then other motor neuron disorders such as spinal muscular atrophy with respiratory distress, X-linked spinal muscular atrophy, distal spinal muscular atrophy, and juvenile amyotrophic lateral sclerosis should be considered. If EMG, nerve conduction study, and repetitive stimulation reveal characteristic patterns associated with diseases in muscle, nerve, or neuromuscular junction, then further diagnostic tests, including muscle or nerve biopsy and edrophonium test, may be performed. When disease of the neuromuscular system is ruled out, then one should pursue diagnostic tests to identify spinal cord or brain anomalies by imaging studies such as magnetic resonance imaging or computed tomography scans. Other diagnostic tests should then be performed to identify systemic diseases, such as metabolic disorders or other genetic disorders.

Clinical Management of Newly Diagnosed Spinal Muscular Atrophy Patients

Many care issues arise when a patient is newly diagnosed with spinal muscular atrophy. Clinicians need to address the various aspects of care issues as soon as possible.

Family Education and Counseling

Because of the complexity of medical problems associated with the diagnosis of spinal muscular atrophy, the committee suggests that medical providers designate a person to meet with the family. This person is usually a pediatric neurologist or a geneticist. The primary care physician (pediatrician or family physician) should be well informed of the multidisciplinary needs of these patients and play a central role in coordinating follow-up care. During the first meeting with parents, it is important to explain the disease process, pathogenesis, phenotype classification, and the patient's prognosis. The physician should also formulate a plan of multidisciplinary intervention with the family. This usually includes referral to a pediatric neuromuscular clinic and/or pediatric subspecialties such as genetics, pulmonary, gastroenterology/nutrition, and orthopedic/rehabilitation. The families will appreciate online resources for further information regarding spinal muscular atrophy. Providing information on spinal muscular atrophy patient advocacy groups has proved to be the most useful to help families cope with the diagnosis (please see the acknowledgments section for links to some patient support group Web sites). Several clinical trials are currently in progress both in the

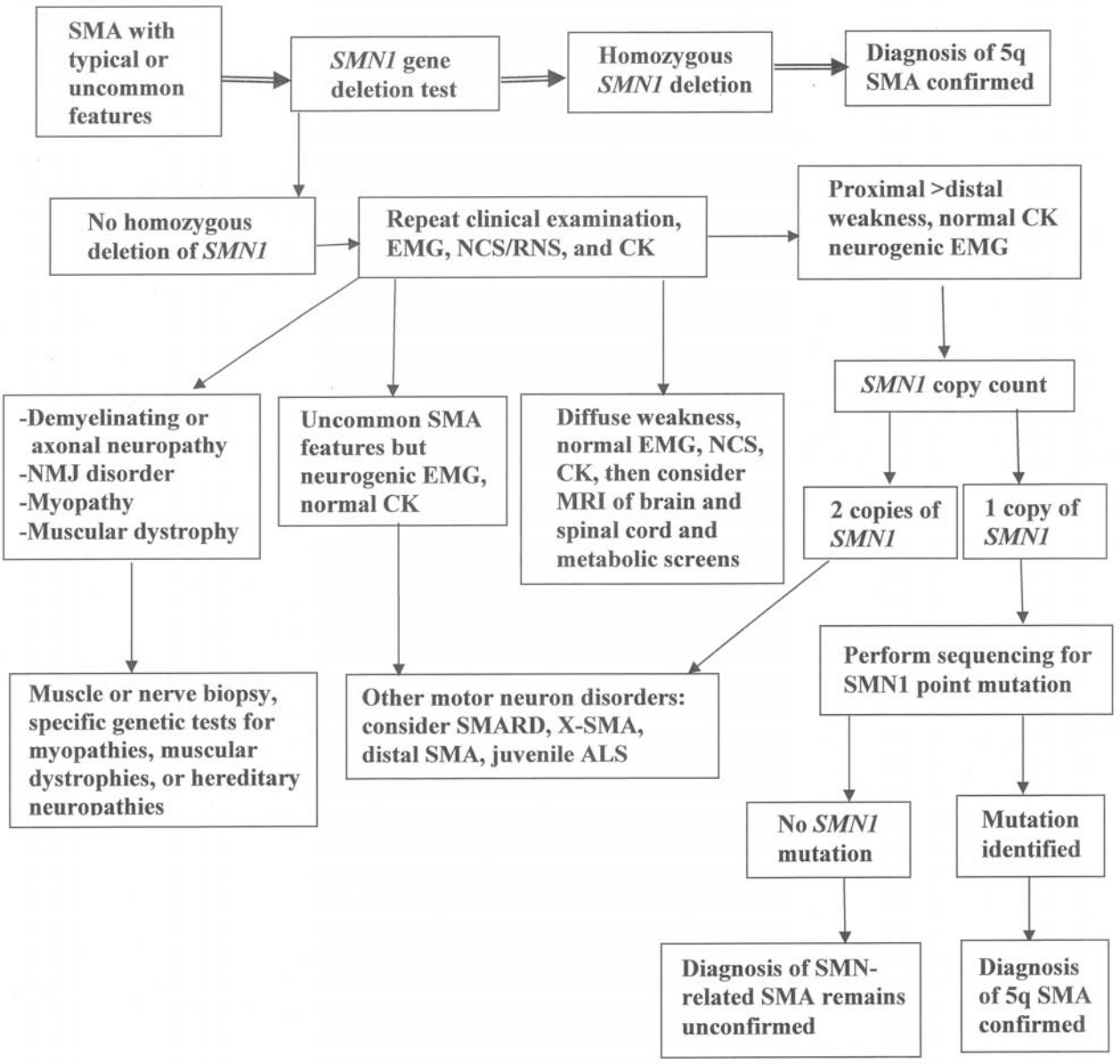

Figure 1. Diagnostic evaluation for spinal muscular atrophy. A diagnostic algorithm for spinal muscular atrophy and other neuromuscular disorders. The Standard of Care Committee recommends the stepwise diagnostic procedure outlined in this flow chart when encountering patients with clinical symptoms of spinal muscular atrophy (SMA). Please see text for detailed explanation of this diagram. EMG = electromyography; NCS = nerve conduction study; RNS = repetitive nerve stimulation; CK = creatine kinase; NMJ = neuromuscular junction; MRI = magnetic resonance imaging; SMARD = spinal muscular atrophy with respiratory distress; X-SMA = X-link SMA; ALS = amyotrophic lateral sclerosis.

United States and in Europe. Physicians should provide information regarding these trials or refer families to clinical trial Web sites (www.clinicaltrials.gov provides a current listing of open clinical trials). Many factors can influence the families' choice to participate in a clinical trial. The families should be encouraged to contact as many study sites as possible before they decide to participate in any trial.

Genetic Topics

Several genetic topics should be addressed with the diagnosis of spinal muscular atrophy. This is often done by a neurologist or geneticist. Topics related to the genetics of spinal muscular atrophy, such as autosomal recessive inheritance and genomic structure of *SMN—SMN1* and *SMN2*

copies—should be explained to the family. The current literature suggests *SMN2* copy numbers correlate with spinal muscular atrophy clinical phenotypes.[24-27] However, although a higher copy number of *SMN2* is correlated with milder phenotype, phenotypes can vary substantially given *SMN2* copy number. Therefore, predicting clinical phenotype using *SMN2* copy number can be risky and is not currently recommended. Other important genetic topics include sibling recurrence risk, carrier testing, and information that may help with reproductive planning (prenatal diagnosis or preimplantational diagnosis). Presymptomatic diagnosis of unaffected siblings is controversial. According to American Society of Human Genetics guidelines, presymptomatic diagnosis in children should be considered only if early intervention can delay the onset or slow the progression of the disease.[28] The committee agrees that presymptomatic diagnosis of at-risk siblings of spinal muscular atrophy patients may lead to early intervention and improve clinical outcome. Therefore, parental request of testing unaffected siblings of the spinal muscular atrophy patient should be granted. The current *SMN1* deletion test will detect the *SMN1* copy number and provide the information of whether the sibling is affected (0 copy) or is a carrier (1 copy). The topic of neonatal screening is also controversial. Although there is currently no proven therapy in spinal muscular atrophy, the committee recognizes the utility of neonatal screening as a tool for identifying effective treatments. Furthermore, in view of recent therapeutic advances, it is possible that in the future, spinal muscular atrophy may be treated more effectively if presymptomatic patients are detected through neonatal screening and treatment is started prior to weakness becoming apparent.

Consensus on Pulmonary Care

Overview of Pulmonary Problems in Spinal Muscular Atrophy

The key respiratory problems in spinal muscular atrophy are as follows:

1. impaired cough resulting in poor clearance of lower airway secretions;
2. hypoventilation during sleep;
3. chest wall and lung underdevelopment; and
4. recurrent infections that exacerbate muscle weakness.

Pulmonary disease is the major cause of morbidity and mortality in spinal muscular atrophy types 1 and 2 and may occur in a small proportion of patients with spinal muscular atrophy type 3. Without respiratory support, infants who are unable to sit usually die before the age of 2 years.[8] Pulmonary compromise is caused by a combination of inspiratory and expiratory muscle weakness, with greater involvement of expiratory and intercostal muscles. The diaphragm is relatively spared. In nonsitters, the result is a bell-shaped chest with sternal depression. In older sitters and walkers, respiratory function may be compromised further by scoliosis. Swallowing dysfunction and reflux are important contributors to pulmonary morbidity. Individuals tend to progress to daytime respiratory failure via a sequence of recurrent chest infections, nocturnal oxygen desaturation, nocturnal hypoventilation, and then daytime hypercarbia.[29-31] In contrast to Duchenne muscular dystrophy, there is no strong correlation between pulmonary functional score and need for mechanical ventilation in spinal muscular atrophy.[32] However, baseline assessment and longitudinal monitoring can identify those at risk for sleep-disordered breathing and ineffective clearance of secretions. There are several case series of the natural history of severe to mildly affected spinal muscular atrophy patients but no large prospective study of treatment intervention.[9,33] The evidence base is limited by heterogeneous groups of patients with a mixture of neuromuscular disorders in natural history and intervention studies, variable classification of spinal muscular atrophy subtypes, and different respiratory support protocols employing a range of ventilators and cough assistance techniques.[30,33-36] Case series[29,30,36,37] and consensus conference[38,39] evidence demonstrate that ventilatory support should be added at night if sleep-disordered breathing is present and cough assistance provided if cough efficiency is reduced. Figure 2 shows a flow chart for pulmonary natural history, assessment, and intervention in spinal muscular atrophy.

Assessment and Monitoring

There is no formal study evaluating any protocol for routine pulmonary assessment of patients with spinal muscular atrophy. However, consensus was achieved within the pulmonary working group on current standard of care for spinal muscular atrophy. The following assessments should be used during baseline and subsequent evaluations of respiratory status and are listed by order of importance as identified by the Delphi survey. Assessment frequency depends on the clinical status and rate of progression of disease for each individual. Suggested frequency of evaluation is every 3 to 6 months, less often in stable walkers, and more frequently in clinically unstable nonsitters.

Nonsitters

Recommendations for respiratory assessment include evaluation of cough effectiveness, observation of breathing, and monitoring gas exchange. Respiratory muscle function tests are indirect measures of cough effectiveness and include peak cough flow, maximal inspiratory pressure, and maximal expiratory pressure. The majority of nonsitters with spinal muscular atrophy may be too weak or too young to perform

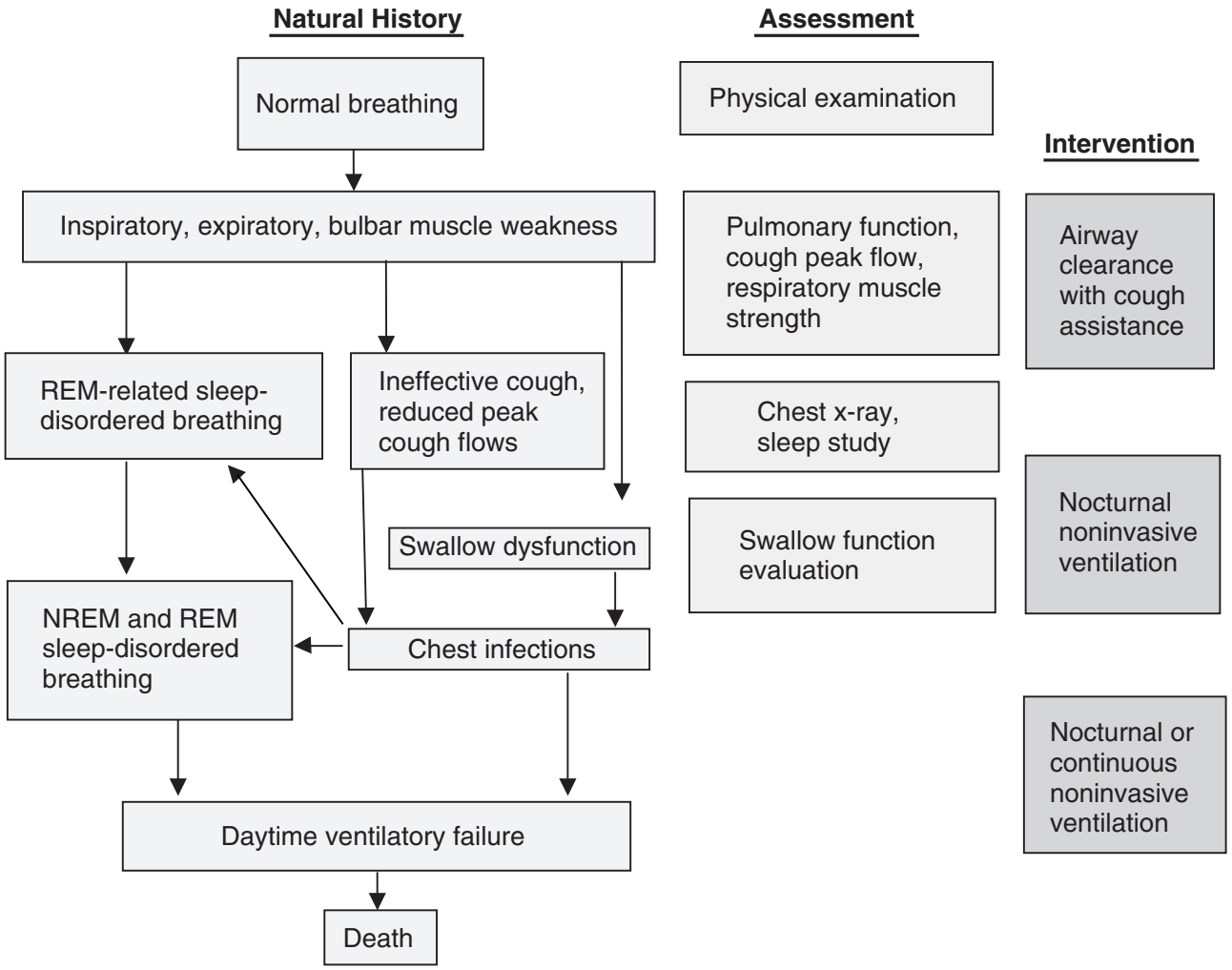

Figure 2. Summary of the natural history of pulmonary problems, assessment, and intervention in spinal muscular atrophy. The progression of pulmonary problems is accompanied by appropriate assessment and intervention strategies. REM = rapid eye movement; NREM = non–rapid eye movement.

pulmonary function testing. Therefore, the most useful evaluation of respiratory muscle function may be observation of cough ability. The physical examination also provides an important assessment of respiratory status including respiratory rate, work of breathing, presence of paradoxical breathing, chest wall shape, and skin color (cyanosis or pallor). Gas exchange monitoring, including pulse oximetry, can be used as a spot check during the day for hypoxemia and as a guide to direct airway clearance. For example, if oxygen saturation is less than 94%, airway clearance techniques should be used. Overnight pulse oximetry with chart recording can be used to screen for nocturnal hypoxemia. Routine overnight monitoring using pulse oximetry may help identify unsuspected hypoxic events but is usually very disruptive to the family due to frequent false alarms. Currently there are no data to support routine continuous oximetry monitoring. Further research is needed before recommending this as part of routine clinical care.

End-tidal carbon dioxide, transcutaneous CO_2, and serum bicarbonate measurement were also identified as important assessment tools. However, serum bicarbonate may give a false sense of reassurance, as normal values may exist despite significant respiratory compromise during sleep. End-tidal carbon dioxide and transcutaneous CO_2 are frequently difficult to obtain and not available routinely. If available, these measurements can be used to assess for sleep-related hypoventilation. The onset of hypoventilation is insidious, and patients may be clinically asymptomatic. Initially hypoventilation will occur in sleep (particularly rapid eye movement sleep), but as deterioration progresses, daytime respiratory function will be impacted.[29,31] Polysomnography is a diagnostic tool during which respiration and sleep state are continuously monitored,[40] and thus it identifies the presence and severity of sleep-disordered breathing.[41] Polysomnography is useful in nonsitters, even in children without obvious symptoms, and can be used to initiate and

titrate respiratory support. When polysomnography is not available, an alternative is to use a 4-channel sleep study that records end-tidal carbon dioxide or transcutaneous CO_2, oxygen saturation, heart rate, nasal airflow, and chest wall movement during sleep. In cases where neither polysomnography nor 4-channel study is available, overnight pulse oximetry with continuous CO_2 monitoring may provide useful information about nighttime gas exchange. However, this will not detect sleep-disordered breathing not associated with oxygen desaturation or CO_2 retention. Further study to better identify the optimal methods for evaluation and monitoring is recommended. Additional screening tests include a baseline chest x-ray to provide an initial reference point and for comparison during respiratory deterioration or unexplained hypoxemia due to unsuspected atelectasis. Although formal radiologic evaluation of swallowing was not ranked very highly for routine evaluation during this Delphi survey, the risk for dysphagia and aspiration is high in nonsitters. Therefore, formal evaluation of swallowing is indicated in cases of acute unexplained respiratory deterioration and recurring pneumonia. Arterial blood gases for routine monitoring of respiratory function are not recommended because the discomfort could result in apnea or spurious hyperventilation.

Sitters

Recommendations of respiratory assessment for sitters are similar to nonsitters and include physical examination and evaluation of cough effectiveness with respiratory muscle function tests (maximal inspiratory pressure, maximal expiratory pressure, and peak cough flow) as described above. In addition, sitters should be evaluated for presence and severity of scoliosis and consider further evaluation with radiographs. Additional recommended assessments include forced vital capacity and lung volume measurements during pulmonary function tests, assessment of sleep-disordered breathing, and pulse oximetry monitoring. Less important assessments identified for sitters include blood gas, CO_2 monitoring, and chest x-ray. Routine swallow study was not recommended for sitters unless clinically indicated.

Walkers

In general, spinal muscular atrophy walkers have relatively preserved pulmonary function until late into their disease course. Recommendations for routine assessment include complete pulmonary function tests, including spirometry, lung volumes, and respiratory muscle function tests. In addition, cough effectiveness and the physical examination are important routine assessments. Further evaluation should be directed by clinical symptoms and indications.

Anticipatory Respiratory Care

Providing families with information about options for care and anticipating future needs are crucial to respiratory management of spinal muscular atrophy. Nonsitters are the most fragile group, and early discussions should include the option of noninvasive ventilation and secretion management because of the rapid progression of the disease. Ongoing discussion of the family's desires for support should occur, and the result should be a negotiated care plan with maximums and minimums outlined.[42]

In addition, anticipatory guidance and education for chronic care, illness management, and perioperative care should be provided. Day-to-day management should include understanding the child's baseline and deviations from his or her baseline, routine cough and secretion management techniques, understanding hypoventilation, and intervention. Illness management includes rapid access to specialty medical care providers, airway clearance and secretion management techniques, respiratory support (including noninvasive ventilation), nutrition and hydration management, and a low threshold to start antibiotics. Routine immunizations, including influenza vaccine, pneumococcus vaccine, and respiratory syncytial virus prophylaxis (palivizumab), are recommended.

Chronic Management

Essential to chronic management is discussion of the family's goals, which includes balancing caring for the child at home for as long as possible, long-term survival, quality of life and comfort, and the availability of resources. Goals of chronic management are to normalize gas exchange, improve sleep quality, facilitate home care, reduce hospitalizations and intensive care unit care, and reduce the burden of illness on the family. There is insufficient evidence, but based on experience and consensus, early aggressive and proactive intervention may prolong life without compromising quality of life.

Airway Clearance

Airway clearance is very important in both acute and chronic management of all patients with spinal muscular atrophy. Caregivers of these patients should learn to assist coughing in all patients with ineffective cough. These techniques include manually and mechanically assisted cough.[43-46] Availability of mechanically assisted cough devices (mechanical insufflation-exsufflation) varies by country but is now widely accepted in management of neuromuscular disease in the United States.[47] Daily assisted cough is recommended in more severely affected patients. Secretion mobilization techniques are also helpful and include chest physiotherapy and postural drainage. Oximetry should be used to provide feedback to guide therapy. Oral suctioning can assist in secretion management after assisted coughing. There is no evidence to support specific secretion mobilization devices such as high-frequency chest wall oscillation and intrapulmonary percussive ventilation in the spinal muscular atrophy population for chronic management.

Respiratory Support

In patients with daytime hypercapnia, respiratory support is clearly indicated. In children with sleep-disordered breathing, nocturnal noninvasive ventilation reduces symptoms of sleep disturbance, nocturnal sweating, and morning headaches and improves appetite and concentration.[29] Objectively, noninvasive ventilation reduces respiratory disturbance index, improves sleep stage distribution,[30] and enhances quality of life. In a randomized controlled trial using mixed groups of patients with neuromuscular disease who showed nocturnal hypoventilation and daytime normocapnia, noninvasive ventilation significantly improved nocturnal blood gas tensions.[48] Noninvasive ventilation with bilevel positive pressure support has been studied most frequently, although there is no evidence to suggest any 1 type of ventilator interface is superior. In addition, the optimal settings for noninvasive ventilation have not been established. In general, noninvasive ventilation settings are individualized to achieve adequate inspiratory chest wall expansion and air entry and normalization of oxygen saturation and end-tidal carbon dioxide or transcutaneous CO_2 measurements. Noninvasive ventilation should be combined with airway clearance techniques.

In nonsitters, care without ventilation support is an option if the burden of treatment outweighs benefit. Noninvasive ventilation can be used palliatively to facilitate discharge to home from a hospital and reduce work of breathing. Continuous positive airway pressure may be an option in a very young nonsitter infant who is not synchronous with bilevel positive airway pressure and can be used with the goal of transitioning to bilevel positive airway pressure. Use of noninvasive ventilation with high-span bilevel positive airway pressure, even for short daytime periods, may improve chest wall and lung development and reduce ribcage and sternal deformity in nonsitters and sitters,[49] resulting in potential beneficial effects on pulmonary function. Uncommonly, adult walkers may develop sleep-disordered breathing or acute ventilatory failure at the time of a chest infection or intercurrent event (eg, surgery). Noninvasive ventilation is an appropriate intervention and may be required during sleep chronically.

Tracheotomy for chronic ventilation is a decision that needs to be carefully discussed if requested by parents. In nonsitters, this is controversial and an ethical dilemma. There is a large spectrum of options that can be provided, ranging from no respiratory support to noninvasive ventilation to tracheotomy and mechanical ventilation. Our recommendation is to explore options with the family regarding the child's potential, quality-of-life issues, and family's desires.[50] Palliative care is an option for nonsitters. It should be noted that noninvasive ventilation can be used as a routine therapy or as a palliative tool. A key goal is to prevent pediatric intensive care unit stays and avoid tracheotomy if possible. If supportive ventilation is chosen by the family, noninvasive ventilation is recommended.

Additional Management

Recommended additional therapies are routine vaccinations, appropriate nutritional support orally or via a feeding tube, hydration management, and medical or surgical gastroesophageal reflux disease management. In addition, medical management for saliva control may be considered. Inhaled bronchodilators should be considered in children with asthma or bronchial hyperresponsiveness. Use of these agents in other situations requires further evaluation. There is no evidence to support the use of mucolytics on a chronic basis.

Perioperative Care

Patients with spinal muscular atrophy are at high risk for postanesthesia complications, which may lead to prolonged intubation, nosocomial infections, tracheotomy, and death. Perioperative complications include upper airway obstruction, hypoventilation, and atelectasis from impaired cough and impaired mucociliary clearance due to anesthetic agents. Postoperative pain may exacerbate respiratory compromise. Noninvasive ventilation associated with aggressive airway clearance techniques can successfully treat hypoventilation and airway secretion retention.

It is crucial that the patient's respiratory status be optimized before surgery. Preoperative evaluation, including pulmonary consultation, is strongly recommended. The assessment of respiratory function should include a physical examination, measurements of respiratory function and cough effectiveness, chest x-ray, and, if at risk, an evaluation for sleep-disordered breathing. In addition, complicating factors should be considered, including oropharyngeal aspiration, gastroesophageal reflux, and asthma. If measurements of respiratory function and/or sleep study are abnormal, nocturnal noninvasive ventilation and assisted coughing techniques may be indicated before surgery. The patient should become familiar with these techniques prior to surgery. The anesthesiology preoperative evaluation should include assessment for possible difficult intubation due to jaw ankylosis. If present, intubation should be performed by fiberoptic bronchoscopy.

Postoperative management should be determined by preoperative respiratory function and the type of surgery performed. Patients with normal cough clearance and relatively preserved muscle function are not at an increased risk for postoperative complications. Patients with decreased respiratory muscle strength require close monitoring and aggressive respiratory management. Any patient who requires respiratory support during sleep will require similar respiratory support in the immediate postoperative course. Extubation in the recovery room to noninvasive ventilation should be planned as a bridge to weaning to the patient's baseline respiratory support. Careful planning and coordination with the hospital respiratory therapists are crucial for success in this setting. Patients with continuous ventilator

support requirements (either via noninvasive interface or via tracheotomy tube) or patients who receive muscular blocking agents during surgery are best transferred directly from the operating room to the intensive care unit. Patients are encouraged to bring their personal devices, such as noninvasive ventilation and mechanical insufflation-exsufflation-E machines, to use in the postoperative period because the availability of these devices in hospitals may be limited. Although oxygen is used frequently in the postoperative setting, it must be applied with caution in the patient with spinal muscular atrophy. Hypoxemia secondary to hypoventilation may be mistaken with hypoxemia due to other causes, such as mucus plugging and atelectasis. End-tidal carbon dioxide or transcutaneous CO_2 monitoring or arterial blood gas analysis will facilitate appropriate oxygen use. Adequate pain control will aid in preventing hypoventilation secondary to splinting. Postoperative pain management should be titrated to promote airway clearance and minimize respiratory suppression. Transient increased respiratory support may be needed while controlling postoperative pain.

Acute Care Management

The goal of acute management is to normalize gas exchange by reducing atelectasis and enhancing airway clearance where possible by noninvasive respiratory support. Blood gas monitoring may be of benefit.

Airway Clearance

For nonsitters, sitters, and walkers experiencing acute illness, airway clearance with manual cough assist or mechanical insufflation-exsufflation, together with oral or airway suctioning, chest physiotherapy, oximetry feedback to guide airway clearance, and postural drainage, are important and recommended. Assisted cough techniques are preferred over deep suctioning and bronchoscopy.

Respiratory Support

Nonsitters. In acute illness, a vicious cycle of added ventilatory load, increased respiratory muscle weakness, and ineffective secretion clearance leads to ventilatory decompensation. Acute use of noninvasive ventilation reverses these features. Continuous positive airway pressure is not indicated in this situation because it does not reduce the ventilatory load. In nonsitters and sitters already using nocturnal noninvasive ventilation, daytime use may be required during acute illness, and airway clearance techniques can be carried out during noninvasive ventilation. Noninvasive ventilation in combination with airway clearance techniques may reduce the need for intubation. Oxygen therapy entrained into the noninvasive ventilation circuit should be used to correct oxygen desaturation, after inspiratory and expiratory positive pressure settings are optimized and airway clearance techniques are optimally utilized. If a noninvasive approach fails, nonsitters can be intubated and mechanically ventilated as a short-term measure. After recovery from the acute illness and arterial oxygen saturation on room air has normalized, they should be extubated back to noninvasive ventilation. Decision making about escalation to intubation should be carried out in advance as part of anticipatory care planning. In nonsitters with increasingly frequent acute pulmonary infections, tracheotomy and ventilation can be considered but may not improve quality of life or reduce hospitalizations. A tracheotomy is not an acute intervention. A noninvasive approach is preferred where feasible. In some nonsitters, with deteriorating function, it may be appropriate to redirect care to a palliative approach.

Sitters. For those already using nocturnal noninvasive ventilation, daytime noninvasive ventilation may be needed during acute illness. Noninvasive ventilation in combination with airway clearance techniques may reduce the need for intubation. Oxygen therapy and need for transient intubation should be carried out as outlined above for nonsitters. Continuous positive airway pressure and/or a tracheotomy are not appropriate interventions in sitters.

Walkers. Walkers may need noninvasive ventilation during an acute illness. Noninvasive ventilation in combination with airway clearance techniques may reduce the need for intubation. Oxygen therapy and need for transient intubation should be carried out as outlined above for nonsitters. If noninvasive ventilation was needed during an acute illness, noninvasive ventilation should be considered for home use. Continuous positive airway pressure and/or a tracheotomy are not appropriate interventions in walkers.

Additional Management

For nonsitters, sitters, and walkers, recommended additional therapies are antibiotics, adequate nutritional support via nasogastric or nasojejunal or gastrostomy tube, hydration, and gastroesophageal reflux management (see more details in the next section). In patients with bronchial hyperresponsiveness or asthma, bronchodilator therapy and inhaled steroids may be indicated. Uses of inhaled mucolytics, bronchodilators, and corticosteroids are areas in need of further research.

Conclusion

Pulmonary disease is the major cause of morbidity and mortality in spinal muscular atrophy types 1 and 2. Respiratory muscle weakness results in impaired cough and ability to clear lower airway secretions, lung and chest wall underdevelopment, and hypoventilation. Respiratory care of patients with spinal muscular atrophy is essential to their survival and quality of life. The pulmonary working group has achieved the following consensus recommendations:

1. Referral for respiratory care evaluation and discussion of options should occur shortly after diagnosis. Key components of the respiratory assessment include evaluation

of cough effectiveness, observation of breathing, and monitoring gas exchange.
2. Chronic respiratory management includes providing methods for airway clearance, including mechanical insufflation-exsufflation or manual cough assist and noninvasive ventilatory support. Routine immunizations are also recommended.
3. Discussion with families about the options for respiratory care and identifying the goals for chronic and acute respiratory care should occur early in the disease course and continue in an ongoing dialogue.
4. Acute respiratory illness management requires increased airway clearance and secretion management techniques using mechanical insufflation-exsufflation or manual cough assist, increased respiratory support (including noninvasive ventilation), nutrition and hydration management, and a low threshold to start antibiotics.
5. Perioperative care includes a thorough preoperative evaluation of respiratory status, ideally by a pulmonologist, and anticipatory guidance of the surgical team and postoperative management team regarding optimal care.

Future Research Directions

The following topics were identified as areas in which no consensus could be achieved because research data are lacking. These areas are in need of further study:

1. Optimal methods for evaluation and monitoring of hypoventilation.
2. Use of pulse oximetry in the home.
3. Optimal secretion mobilization techniques (eg, chest physiotherapy, postural drainage, high-frequency chest wall oscillation, and intrapulmonary percussive ventilation).
4. Optimal ventilatory support settings.
5. Effectiveness of inhaled and nebulized medications, including bronchodilators, mucolytics, and corticosteroids.

Consensus on Gastrointestinal and Nutritional Care

Overview of Gastrointestinal and Nutritional Complications in Spinal Muscular Atrophy

The key clinical problems associated with gastrointestinal and nutritional complications in spinal muscular atrophy are as follows:

1. *Feeding and swallowing problems*. Bulbar dysfunction is universal in spinal muscular atrophy patients with severe weakness and can result in feeding and swallowing difficulties and aspiration pneumonia, which often results in death. The severity of bulbar dysfunction is variable in patients with spinal muscular atrophy of intermediate severity and rare in those who are mildly affected.
2. *Gastrointestinal dysfunction*. Gastroesophageal dysmotility problems include constipation, delayed gastric emptying, and potentially life-threatening gastroesophageal reflux.
3. *Growth and undernutrition/overnutrition problems*. Without optimal management, growth failure is universal in nonsitters, whereas excessive weight gain is more common in sitters and walkers.
4. *Respiratory problems*. The presence of respiratory complications (weak cough, increased work of breathing, dyspnea, pneumonias, and cyanosis or desaturation with feeds) raises concern for gastrointestinal problems of aspiration and gastroesophageal reflux, which can be serious and life-threatening. Increased work of breathing may also result in increased energy expenditure.

In the following sections, we will take a problem-oriented approach to discussing the evaluation and management of these problems during chronic care and acute illness.

Feeding and Swallowing Problems

Feeding and swallowing difficulties are common in nonsitters and sitters but are rarely a concern in walkers. Key symptoms of feeding difficulties include prolonged mealtime, fatigue with oral feeding, and evident choking or coughing during or after swallowing. The presence of recurrent pneumonias is a potential indicator of aspiration, which may be silent (ie, without evident choking or coughing). Articles in the literature addressing the role of oral motor structures and specific chewing and swallowing impairments that impact oral feeding performance in spinal muscular atrophy are limited to class III and IV evidence. One review found a 36% prevalence of at least 1 feeding-related issue in children with spinal muscular atrophy.[51] Several other population studies and case series report swallowing problems in patients with spinal muscular atrophy.[51-53] Difficulties in the preoral phase included limited mouth opening and difficulties in getting food to the mouth for self-feeding.[51] In the oral phase, difficulties included weak bite force, reduced range of mandibular motion limiting mouth opening, and increased fatigue of masticatory muscles.[54] This affects biting and chewing abilities and can lead to prolonged mealtimes and fatigue, precluding sufficient intake. Masticatory and facial muscle weakness affects oral bolus control, chewing, and bolus propulsion, all of which contribute to reduced feeding efficiency. Difficulties with strength and efficiency are reported in the oral and pharyngeal phase of the swallow. Poor coordination of the swallow with airway closure can lead to penetration and aspiration of the airway. Poor head control may also be a factor in the development of feeding difficulties, precluding neck tuck or other compensatory postures to enhance the safety of swallowing.[55] The psychosocial impact of feeding difficulties on these children and their family should not be underestimated. Prolonged mealtimes can put time pressure on other activities. In addition, their inability to feed themselves can make these children seem more dependent

than their peers and lead to a sense of loss of control. In the weakest children, tube feeding can limit the nurturing role parents perceive from being able to orally feed their child.[53]

Evaluation of Feeding and Swallowing Problems

Assessment of feeding problems should be performed by a feeding specialist, most commonly a speech or occupational therapist. Routine clinical evaluation of feeding and swallowing difficulties should include a feeding assessment. Videofluoroscopic swallow studies should be performed when indicated. A feeding case history with mealtime observation is desirable. Examination of oral structures that influence feeding efficiency and consideration of the effect of positioning and head control on feeding and swallowing are essential. Videofluoroscopic swallow studies should be carried out after initial assessment if there are concerns about swallow safety.[51,53,56,57] Laryngeal aspiration requires specific assessment, as it is sometimes silent (ie, no clearing cough is triggered).[56] In severely affected children, vocal fold paralysis and consequent inability to protect the airway may be a diagnostic sign.[57,58] A videofluoroscopic swallow study is not simply a diagnostic test of aspiration but is an opportunity to evaluate therapeutic strategies, such as adapted food texture and positioning, to assess impact on swallow function. As position and consistency can affect swallow physiology, it is important that the videofluoroscopic swallow studies procedure is as representative of the child's usual meal and feeding position as possible.

Management of Feeding and Swallowing Difficulties

Treatment should aim at reducing the risk of aspiration during swallow and optimizing efficiency of feeding and promote enjoyable mealtimes. A Cochrane review[59] of treatment of swallowing difficulties in chronic muscle disease concluded it was not possible to determine the benefit or otherwise of dietary and feeding advice, surgical intervention (cricopharyngeal myotomy or upper esophageal dilatation), and enteral feeding. Changing food consistency and optimizing oral intake are appropriate treatment strategies. The literature suggests there is currently widespread use of consistency modification in helping to optimize oral intake.[51,53] A semisolid diet can be used to compensate for poor chewing and reduce length of mealtimes. Thickened liquids may protect against aspiration of thin fluids. Preferably, this intervention would be evaluated objectively on videofluoroscopic swallow studies. In 1 study, complete restriction orally to eliminate risk of aspiration during swallowing was not found to significantly affect the clinical course in severe spinal muscular atrophy.[60] This study failed to consider the risk of aspiration due to concomitant gastroesophageal reflux. Positioning and seating alterations and orthotic devices (eg, Neater Eater, elbow support, valved straw) to enhance self-feeding ability may improve swallow safety and efficiency.[51] Such interventions should be planned in liaison with an occupational therapist and/or physiotherapist as required. There is currently no supporting evidence that oral motor treatment programs impact safety or efficiency of oral feeding.

The gastrointestinal/nutrition working group did not reach consensus regarding when to refer a patient with spinal muscular atrophy for consideration for gastrostomy tube placement and whether one should supplement or replace oral feeding with tube feeding in a nonsymptomatic patient. Some practitioners prefer a proactive approach, particularly in the nonsitters, whereas others believe that exposing such patients to the risk of surgery is inappropriate prior to the onset of symptoms. However, 1 clear consensus is that optimal management requires proactive nutritional supplementation as soon as inadequate oral intake is recognized. Whether a gastrostomy tube is placed in a particular child often requires extensive discussion with multiple caregivers. It usually takes time to schedule a surgical procedure like gastrostomy tube placement. In the interim, nutritional supplementation via nasogastric or nasojejunal feeding is desirable. Nasojejunal feeding may be preferable in circumstances when gastroesophageal reflux with aspiration is a concern, especially when the patient is on ventilatory support. However, technical difficulty may prevent its feasibility. Gastrostomy tube feeding is the optimal method of feeding when insufficient caloric intake or unsafe oral feeding is of concern. It prevents the potential morbidity associated with prolonged use of either nasogastric or nasojejunal tubes. The presence of a nasojejunal or nasogastric tube may also result in a less-than-ideal mask fit when there is a need for the use of noninvasive ventilation such as bilevel positive airway pressure.

There are several options for gastrostomy tube placement, including insertion via percutaneous methods with endoscopic guidance, or placement via open or laparoscopic surgical techniques[61] together with an antireflux procedure such as Nissen fundoplication. The open surgical technique is associated with a relatively large upper abdominal incision, increased postsurgical pain, and risk for respiratory complications due to diaphragmatic splinting. A laparoscopic surgical technique provides the best possible setting for immediate or early postoperative extubation.[62] Such procedures are typically performed with general anesthesia, although placement using percutaneous methods with endoscopic guidance is performed in some centers with conscious sedation and local anesthesia. Care should be taken to minimize the amount of fasting preoperatively and to resume full nutritional support as quickly as possible following the procedure. Possible pulmonary complications of sedation should be anticipated and may require treatment with noninvasive ventilation (see "Pulmonary Care").

Gastrointestinal Dysfunction

Children with spinal muscular atrophy suffer from the following gastrointestinal problems: gastroesophageal

reflux, constipation, and abdominal distension and bloating. Gastroesophageal reflux is an important determinant of mortality and morbidity in patients with spinal muscular atrophy. It can be associated with silent aspiration and results in pneumonias and, at times, life-threatening events.[60] Frequent "spitting up" or vomiting after meals, complaints of chest or abdominal discomfort, bad breath, or obvious regurgitation of feeds may indicate gastroesophageal reflux. Some children may refuse feeds when they develop discomfort with swallowing, placing them at risk for undernutrition. High-fat foods delay gastric emptying and increase the risk of gastroesophageal reflux. Constipation is a frequently reported problem and is likely multifactorial in origin (ie, abnormal gastrointestinal motility, reduced intake of dietary fiber, inadequate fluid intake, low muscle tone of the abdominal wall). Infrequent bowel movements can lead to abdominal distention and bloating. In children dependent on their abdominal muscles to assist with respiration, desaturation or respiratory distress in association with attempted bowel movements may occur.

Evaluation of Gastrointestinal Dysfunction

The symptoms of gastroesophageal reflux (emesis, regurgitation, gurgling after feeds) should be sought early. A routine upper gastrointestinal series is recommended for presurgical evaluation for gastrostomy tube placement to primarily rule out anatomic anomalies and secondarily to document reflux. In rare cases, esophageal stricture, foreign body, or other abnormality may contribute to swallowing difficulties or gastrointestinal dysmotility. Motility studies, including scintigraphy, can be helpful in documenting delayed gastric emptying, which may contribute to gastroesophageal reflux and early satiety. There are no data to support the routine diagnostic use of pH probe studies in documenting reflux.

Management of Gastroesophageal Reflux

Medical management of gastroesophageal reflux typically involves the use of acid neutralizers (eg, magnesium or calcium carbonate) and/or inhibitors of acid secretion. This latter category includes both histamine blockers and proton pump inhibitors (eg, famotidine, ranitidine, omeprazole). Short-term use of these agents is reasonable for symptomatic management. However, increasing evidence suggests that prolonged use of these agents may be associated with a greater risk for gastroenteritis and pneumonia.[63,64] When delayed gastric emptying or diminished motility is present, prokinetic agents may be useful (eg, metaclopramide, erythromycin). Use of probiotics such as acidophilus or lactobacillus to help maintain a healthy gastrointestinal flora, particularly after antibiotic treatment or in the setting of prolonged use of acid inhibitors, is an area deserving further study.

Gastrostomy tube feeding does not ameliorate gastroesophageal reflux. This is of particular concern in nonsitters who are the least able to protect their airway via a triggered cough. Determining whether aspiration has occurred during swallow or as a result of gastroesophageal reflux is often difficult, and sometimes both may contribute. A laparoscopic antireflux procedure (eg, Nissen fundoplication) is commonly performed as a combined procedure during the same general anesthesia for gastrostomy tube insertion. Although some physicians support a proactive combined laparoscopic Nissen and gastrostomy tube procedure in those children with spinal muscular atrophy who are deemed at greatest risk for aspiration, there is as yet no published data nor consensus to support this strategy. However, in the spinal muscular atrophy patient with medically refractory gastroesophageal reflux, and in whom the benefit is deemed to outweigh the associated surgical and anesthetic risks, laparoscopic Nissen fundoplication during gastrostomy tube placement is supported as an appropriate intervention.

Growth and Undernutrition/Overnutrition Problems

Children with spinal muscular atrophy are at risk for growth failure or excessive weight gain. Growth failure is commonly seen in nonsitters and some sitters, whereas obesity is a problem of the stronger sitters and walkers. There are no articles in the literature that specifically address body composition and growth expectations or typical anthropometric measures in children with spinal muscular atrophy. However, data can be extrapolated from literature on patients with spinal cord injury. Individuals with spinal cord injury have been shown to have lower lean tissue and higher percentage body fat than controls. Body mass index significantly underestimates body fat in these patients.[65,66] Children with spinal muscular atrophy may have acceptable fat mass but may plot as underweight based on weight/height criteria due to the decrease in lean body mass.[53] Hence, normal body mass indexes may not represent the ideal weights for children with spinal muscular atrophy. Decreased activity and lean body mass will lead to reduced resting energy expenditure and increased risk of obesity.

Management of Growth and Undernutrition or Overnutrition Problems

Routine history, physical examination, and monitoring of growth velocity measures (growth charts) form the evaluation process to detect signs and symptoms of growth failure or excess. This will influence decisions regarding when and how to intervene. The goal is to maintain each child on his or her own growth velocity. Growth velocity curves (weight, height/length, weight/height) followed over a period of time are, for the most part, the most accurate indicator of nutritional status. Difficulty in obtaining accurate standing height measurements due to contractures or inability to

stand may complicate growth monitoring in these children. Recumbent length, segmental measurements, or arm span may be useful surrogate markers for linear growth in these children. Other methods for monitoring body composition include skinfold measurements, muscle circumference, or bioelectric impedance analysis.[67] Assessment of nutritional intake by a dietitian or other health care provider proficient in nutrition is recommended at each visit. A 3-day dietary record is a simple and accurate tool that can help assess whether nutritional intake is adequate.[53] A 24-hour food recall is a practical method to highlight major nutritional concerns and to obtain information regarding use of any special supplements. Analysis should target adequacy of macronutrient (including fiber intake) as well as micronutrient intake. Currently, there is no indication for increasing or decreasing specific nutrients (ie, protein, fat, or selected vitamins or minerals). Until more specific data are available, nutrient intake should meet the daily recommended intakes for age. Supplements to provide more than the dietary recommended intake for vitamin, mineral, protein, or fat should be discouraged. Although anecdotal benefit with the use of elemental or semi-elemental formulas has been reported by some families and care providers (satisfactory growth and decreased secretions), there is currently insufficient data to make specific recommendations regarding their use. If an elemental formula is used, a dietitian should be involved to help ensure the child does not receive insufficient or excessive amounts of nutrients, to perform laboratory assessments as needed, and to monitor adequate growth.[68] As previously mentioned, with a reduction in lean body mass, calculated body mass index will significantly underestimate body fat.[66] Children with spinal muscular atrophy may have acceptable fat mass but may be perceived as underweight based on weight/height criteria because of their decreased lean body mass. This will result in inappropriate dietary recommendations that could lead to relative obesity.[53] The spinal muscular atrophy patients at risk for obesity should have growth parameters in the lower percentiles for weight/height and body mass index. In any case, each child should be evaluated individually on a routine basis, with the goal of following their established growth curves and avoiding inadequate or excessive intake. There is some evidence that decreased bone mineral density may occur in nonsitters and sitters, resulting in recurrent fractures in a subset of patients.[69] There is preliminary evidence that dual energy x-ray absorptiometry could be a useful technique for estimating lean versus fat mass in spinal muscular atrophy patients.[70-73] However, insufficient data are available at this time to recommend the routine use of dual energy x-ray absorptiometry scans for monitoring bone mineral density or body composition. Instead, the importance of documenting appropriate intake of calcium and vitamin D was emphasized. There is no consensus regarding performing biochemical tests to monitor nutritional status for patients with spinal muscular atrophy. However, consideration should be given to checking prealbumin levels to help assess adequate protein status.[53]

Management of Nutrition in the Acutely Sick Spinal Muscular Atrophy Patient

Spinal muscular atrophy patients are particularly vulnerable to catabolic and fasting states. Patients with severe muscle wasting from any disorder, including spinal muscular atrophy, are more likely to develop hypoglycemia in the setting of fasting.[74,75] A number of case series and individual case reports have documented secondary mitochondrial dysfunction and abnormalities of mitochondrial fatty acid oxidation in spinal muscular atrophy patients.[76-83] Significant abnormalities are most likely in nonsitters and sitters, increasing their vulnerability for metabolic decompensation in the setting of a catabolic state. Thus, it is necessary to avoid prolonged fasting, particularly in the setting of acute illness, in all spinal muscular atrophy patients. Nutritional intake should be optimized to meet full caloric needs within 4 to 6 hours after an admission for acute illness, via enteral feeding, parenteral feeding, or a combined approach as necessary. Prompt postoperative caloric supplementation is recommended to avoid muscle catabolism, particularly in a child with reduced fat store. If enteral intake is not imminent, then intravenous caloric feeding should be considered.

Conclusion and Future Directions

Because nutritional problems associated with spinal muscular atrophy influence the patient's pulmonary status and general well-being, optimal management of these problems by a multidisciplinary or interdisciplinary team of physicians, speech therapists or occupational therapists, dietitians, and pediatric surgeons should greatly improve survival and quality of life.[62]

The following topics were identified as areas in need of further study:

1. Use of elemental formulas to support/refute perceived benefits of optimal growth and decreased oral and airway secretions.
2. Need for a reduced fat intake, in view of the concern for mitochondrial fatty acid oxidation abnormalities.
3. Need for protein supplementation beyond dietary recommended intake, in view of the problem of muscle wasting/atrophy.
4. Need for checking biochemical tests for metabolic/mitochondrial fatty acid abnormalities, in view of the concern for mitochondrial fatty acid oxidation abnormalities.
5. Need to determine body composition and establish growth charts for the population of patients with spinal muscular atrophy to enable optimal growth monitoring in these patients.

Consensus on Orthopedic Care and Rehabilitation

Overview of Orthopedic Care and Rehabilitation Strategies in Spinal Muscular Atrophy

Key Problems

Muscle weakness of varying severity limits motor function of trunk and upper and lower extremities, resulting in contracture formation, spinal deformity, limited mobility and activities of daily living, and increased risk of pain, osteopenia, and fractures.

Key Evaluation Procedures

These include evaluating range of motion, strength, function, seating and mobility, orthotics, radiographs (spine and other joints), and dual energy x-ray absorptiometry. The value of these procedures varies by degree of functional impairment.

Key Interventions

In nonsitters, nutritional support, posture management, seating, contracture and pain management, therapy for activities of daily living and assistive equipment, wheelchairs for mobility, limb orthotics, and developmental therapies are important. In sitters, wheelchair mobility, contracture management, physical therapy, and occupational therapy are of highest value, with strong considerations for spine and limb orthotics and spine surgery. In walkers, the highest emphasis is on provision of physical therapy, occupational therapy, and wheelchair/mobility, although orthotics, scoliosis surgery, and pain management figured prominently. In the United Kingdom and some other European countries, chest physiotherapy is often done by physical therapists.

Literature Review

Rehabilitation and Orthopedic Problems in Spinal Muscular Atrophy

Literature review pertinent to these rehabilitation and orthopedic concerns reflects similar musculoskeletal and functional problems to those presented in "Key Problems".[1,84-86] Hip subluxation is a common comorbidity in patients with spinal muscular atrophy.[87,88] As patients with spinal muscular atrophy age, there is a significantly higher prevalence of kyphoscoliosis, difficulty coughing, joint contractures, and voice/speech problems in types 1 and 2. In type 3, there is also a significantly higher prevalence of fatigue and hypermobility of the hand.[89] Scoliosis develops in more than 50% of children with spinal muscular atrophy, most commonly in nonambulatory children or in those who lose the ability to walk.[90]

Evaluation

Traditional measurements of strength are not possible in severely affected infants and children; thus, emphasis is on observation of function. Evaluation procedures that address rehabilitation/orthopedic concerns include the CHOP-INTEND,[91] the Hammersmith Functional Motor Scale for Spinal Muscular Atrophy,[92,93] the Modified Hammersmith Functional Motor Scale for Spinal Muscular Atrophy,[94] the Gross Motor Function Measure,[95] and the Motor Function Measurement scale for neuromuscular disease.[86,93] Most children with spinal muscular atrophy require help or supervision with bathing and dressing and assistance with mobility. Stairs present a major obstacle.[96] Early and generalized joint contractures and scoliosis correlate with level of motor function and walking with support, rolling by 5 years of age correlates with eventual walking, and inability to roll correlates with severe disease (greater weakness).[97] Muscle strength can be quantified using myometers, videotaped movements,[98] handheld dynamometers,[99] and quantitative muscle testing in children with the type 2 or 3 forms of the disease.[100-102] Flexion contractures, which affect almost half of spinal muscular atrophy patients, are often noted during periods of inactivity and are considered intractable if greater than 45° Activities of daily living are hampered, and contractures are perceived to be associated with disability in about half. Pain increases in frequency and severity over time and correlates with decreased scores on quality-of-life indicators.[103] In all studies of scoliosis, spine radiographs were routinely used for diagnosis. A retrospective review of spinal full-length radiographs revealed a predominance of right-sided thoracic and thoracolumbar curves and left-sided lumbar curves.[104]

Interventions

No studies directly address physical therapy and occupational therapy as general therapies, although a case report documented the ability of a 20-month-old girl with spinal muscular atrophy to learn to operate a power wheelchair independently in 6 weeks and demonstrated developmental gains in all domains of the Batelle Developmental Inventory over the ensuing 6 months.[65] Regarding other interventions, 3 case series discussed the use of knee-ankle-foot orthoses in patients with spinal muscular atrophy. Evans[105] presented 5 cases (3 who had lost the ability to walk) who were treated with serial casting and bracing and were still ambulatory 2 to 5 years later. Granata presented 7 cases and later 12 cases of patients with type 2 spinal muscular atrophy. All were able to stand independently with knee-ankle-foot orthoses, and 7 achieved assisted ambulation. When compared with a historical control group, the treatment group had less scoliosis. There may have been a trend toward greater hip subluxation in the treatment group.[106]

Consensus Recommendations on Evaluation and Treatment by Functional Levels

The natural history of the disorder should be considered along with the results of the examination and the goals of the patient and family in planning treatment. Intervention should address the problems that were identified through a thorough history and examination. On the basis of the literature review, the results of the Delphi survey, and our group conferences, we list recommendations in this care area by the functional levels of these patients.

Nonsitters

These patients present in early infancy to rehabilitation providers with impairments in respiratory function and profound weakness. Limited range of motion, head control, postural control and alignment, and progressive scoliosis are found. There is significant fatigue during and after medical care and with therapies. Weakness leads to varying functional deficits that interfere with caretakers' abilities to perform activities of daily living and that also limit participation in developmental activities and later in school. A multidisciplinary approach to evaluation and management includes a strong partnership between therapists, patients and families, and physicians. Assessments include physical and occupational therapy and speech therapy if swallowing is impaired or if speech production is affected by jaw contractures and inadequate ventilatory support of voice. Play and occupational support should include lightweight toys and assistive technology with variable controls and a myriad of activation systems. Consideration of the patient's primary posture should direct choice of equipment and devices that support function. Upper extremity orthotics to aid in function include the use of mobile arm supports or slings that augment active range of motion and functional abilities. Use of linear elastic elements to balance out the effects of gravity in multiple dimensions can aid those with proximal weakness and improve control of distal function. Upper extremity or hand orthoses should be considered with caution because attempts to correct postural deviations and compensations with an orthosis may result in reduced function. Compensations due to hypermobility and lack of power should not always be discouraged. Splinting to preserve range of motion and prevent pain may be indicated.

Sitters

Weakness, contractures, respiratory dysfunction, and scoliosis characterize the main problems of this group. These impairments contribute to limitations in mobility, endurance, and activities of daily living. Evaluations by physical therapists, occupational therapists, and orthopedic surgeons include measurement of contractures and strength by goniometry, manual muscle testing, or myometry, with judicious use of spine and hip radiographs. Equipment evaluation includes seating and mobility, positioning, and equipment for self-care. The need for assistive technology and adaptive aids should be determined in the context of improved function. Pulmonary evaluation should be conducted, as it pertains to exercise tolerance and endurance. Evaluations for manual and power mobility may be conducted as early as 18 to 24 months of age. Contracture management and exercise are a major focus of treatment, with implementation of a regular stretching and bracing program to preserve flexibility. Serial casting for contractures may improve participation in a standing program and improve tolerance of bracing. Regular exercise should be encouraged to maintain fitness and endurance and might include swimming and adaptive sports. Lightweight ischial weight-bearing knee-ankle-foot orthoses or reciprocal gait orthoses should be considered for standing or assisted ambulation with a walker for patients with sufficient strength. Where this is not possible, a standing frame or mobile stander with ankle-foot orthoses should be considered. Upper extremity orthotics with mobile arm supports or slings augment active range of motion and functional abilities. Assistive technology and other adaptive equipment to enhance independent work and play should be considered.

Walkers

The combination of proximal weakness and impaired balance results in frequent falls. Limitations are found in transitions between the floor, sitting and standing, distance ambulation, changes in terrain, and stair climbing. There are consistent complaints of fatigue with activity. Musculoskeletal deformities and pain are most commonly reported in late childhood and early adolescence, and with their onset, functional limitations become more pronounced. Patients may present acutely for management of fractures or other musculoskeletal injury. Balance and ambulation evaluations include a specific survey of environmental adaptability and access. Evaluation of joint range of motion and spinal alignment as they affect function, comfort, and balance guides more specific orthotic and spinal assessment and x-rays. Physical and occupational therapy assessments to determine appropriate mobility aids, adaptive equipment, assistive technology, and environmental access will allow patients to maintain independence and mobility and to conserve energy. Activities of daily living assessment for equipment and adaptation may improve independence and access to home and community environment. Nonspine x-rays and dual energy x-ray absorptiometry are considered in the event of acute musculoskeletal injuries as a result of overuse, an accident, or a fall. Treatment and interventions should consider goals of the family and/or caretakers and should be problem-driven. Physical therapy consultation helps to maximize safety, endurance, and independence or to prolong ambulation. Orthotics also support functional walking. Wheelchair

mobility for longer distance transportation adds mobility and independence. Walkers appear less likely to develop scoliosis; thus, continued walking should be encouraged. Contracture management and education to maximize joint protection should be a part of any treatment program. Maximum functional activity includes access to leisure, adaptive sport, and play activities. Regular exercise to maintain fitness and stamina should be encouraged and may include swimming, aquatic therapy, horseback riding, and adaptive sports. Weight management with attention to fitness and education about nutrition are necessary. Equipment needs related to activities of daily living and assistive technology and other adaptive equipment may be useful to enhance abilities for independent work and play. Environmental controls and home modifications to allow for safe accessibility and optimal independence should be explored. Driver's education alternatives and consideration of customized driving controls should be part of the overall rehabilitation management of the adult with spinal muscular atrophy.

Orthotics

In selecting and fabricating an orthosis for patients with spinal muscular atrophy, it is important that the orthotist, therapist, and family work together to ensure that the appropriate orthosis is fabricated and allows wearers to meet their functional goal. For patients with spinal muscular atrophy, it is particularly important that the orthotist has a good background and experience in working with patients with neuromuscular disorders. Familiarity with patterns of weakness and compensations allows the orthotist to choose proper materials and to make adaptations that allow for "best" fit and function.

Spinal orthoses may be used for postural support, but there is insufficient evidence to support delayed curve progression. When used, spinal orthoses should be fabricated with an abdominal cutout to allow appropriate diaphragmatic excursion and access to gastrostomy tubes where present.

Orthopedic Surgery

Surgical correction of scoliosis should be considered based on the patient's curve progression, pulmonary function, and bony maturity. Scoliosis surgery in children with prolonged survival provides benefits in sitting balance, endurance, and cosmesis.[105,107-111] Evidence suggests that earlier surgery results in better outcome. Beneficial effects on pulmonary function remain controversial, but the rate of pulmonary decline may be slowed.[112] Intraoperatively, excessive bleeding may occur. Postoperatively, complications include loss of correction, pseudarthrosis, a requirement for prolonged ventilatory support, and chest and wound infections. Careful consideration is warranted for the spinal muscular atrophy patient who is still ambulatory because altered function, balance, and respiration may result in loss of independent walking. Pelvic obliquity may require surgical fixation.[113] Intraoperative neurophysiologic monitoring may detect temporary abnormalities during scoliosis surgery.[114]A survey of patient/parent satisfaction and clinical/functional outcome was sent to 21 patients with spinal muscular atrophy who underwent operations for scoliosis. Of those who returned the surveys, all found benefit from scoliosis surgery regarding cosmesis, quality of life, and overall satisfaction.[115] Although there is a higher rate of hip subluxation in spinal muscular atrophy, few are painful. Surgical reduction and osteotomy are frequently followed by redislocation.[116,117] In most circumstances, this surgery is avoidable. Ankle and foot deformities make conventional shoes difficult to wear, and orthopedic surgeons may consider soft tissue releases at the child and family's request. In walkers, if soft tissue releases are performed, rapid and aggressive physical therapy may improve outcome.

Perioperative Management in Spinal Muscular Atrophy

Perioperative management and the role of rehabilitation should be customized according to the specifications and needs of the patient and family, therapist, and surgeon. In general, preoperative management includes appropriate modification of the individual's environment, a plan for orthotic intervention, and confirmation of timing and modification of orthoses. New wheelchairs or wheelchair modifications of the seat, back, arm, leg, or headrests are likely to be required. One may anticipate increased sitting height after scoliosis surgery, resulting in the need for van modifications. Families need instruction in transfers, including arrangements for a mechanical lift, if necessary. Arrangements for bathing, toileting, and dressing equipment and potential modifications to clothes for ease in donning and doffing over, under, and/or around casts or orthoses are necessary. Two small studies found that noninvasive positive pressure ventilation, 1 with mechanical-assisted cough training prior to surgery, resulted in successful extubation.[118,119] Incentive spirometry practice may be initiated and coordinated with preoperative noninvasive pulmonary supports, such as bilevel positive airway pressure and exsufflator (cough-assist) devices.

Postoperatively, one must confirm timing of appropriate casting and fitting of orthoses, allowed range of motion, and activity and that appropriate adaptive equipment is available. Therapists can ensure appropriate use of incentive spirometry and instruction of nursing staff and family on bed mobility, transfers, dressing, bathing, and toileting. The individual should be mobilized as soon as possible, as allowed by the procedure and surgeon.

Conclusion

Infants and children with spinal muscular atrophy should have appropriate evaluation for their presenting musculoskeletal and functional deficits. Goals of therapy and surgery depend on functional level and the family's wishes. Even young children should be offered independent mobility and activities of daily living, which includes play. Whenever possible, walking should be encouraged with appropriate assistive devices and orthotics. Hip subluxation is rarely painful, and there is a high risk of recurrence despite surgical correction. Spinal orthoses may provide postural support but do not prevent curve progression and may impair respiratory effort. Scoliosis surgery appears to benefit patients who survive beyond 2 years of age when curves are severe and progressive and should be performed while pulmonary function is adequate. Preliminary studies show the benefit of preoperative training with noninvasive ventilation and cough-assist devices. Intraoperative neurophysiologic monitoring detects early neurologic compromise in some and may improve outcome.

Recommendations for Future Direction

The optimal evaluation procedure to assess motor function in very weak infants is evolving, with ongoing research efforts under way. Many questions remain regarding best practices in therapeutic interventions. There is a need for the development of creative technology to improve independent function. Further research is suggested to evaluate the effects of spinal bracing and surgery on function, balance, and respiratory function. The role of bone density evaluation and treatment of osteopenia must be further examined.

Palliative Care Issues

In most circumstances in the course of medical practice, the goal of therapy—to further quality and extent of life—is straightforward. In the case of patients with spinal muscular atrophy, however, the appropriate goal of therapy may not be clear. Some therapies may be perceived as placing quality of life in conflict with duration of life, prolonging suffering rather than relieving the burden of disease. Thus, there is little national or international consensus about the appropriate level of care, and local experience, training, habit, and resource availability appear to have a large effect upon recommendations and ultimately family decisions about interventional support.[120-122] Although not surveyed formally, the committee is aware of a similar broad range of practice regarding appropriate pulmonary, nutritional, orthopedic, and other forms of therapy.

Optimal clinical care for these patients should be mindful of potential conflict of therapeutic goals. This conflict is made more difficult by the need for surrogate decision makers for a dependent infant and the fact that many—including parents, siblings, other relatives, caregivers, payers, and the wider community—will be affected by and thus have some valid interest in care decisions. The committee reached consensus that these conflicts are real and that there is no moral imperative to any therapy. There is, however, a deep responsibility to present care options in an open, fair, and balanced manner.

A choice for or against interventional supportive care is not a single binary choice, nor must it be unchanging with circumstance. There are, however, some interventions that are better done early so as not to constrain later potential assistance. For example, placement of a gastrostomy tube is better done relatively early, when associated risks are lower, to provide more stable and comfortable nutritional support later when feeding is more tenuous. Similarly, it is important to discuss and determine the appropriate response to potential life-threatening respiratory insufficiency, as emergency resuscitation and endotracheal intubation during times of crisis without prior respiratory support are associated with many more problems in care than when decisions are made in advance. If appropriate, other forms of noninvasive respiratory device that might reduce the potential for emergent respiratory support should be introduced according to increasing need. Whenever possible, caregivers should ideally permit sufficient time after diagnosis prior to discussing these difficult issues; in all cases, sufficient time, honest appraisal of the choices, openness to revisiting decisions made, and personal rapport are essential to these discussions. If appropriate, other family members or trusted friends or spiritual advisors should be invited. End-of-life care decisions need to be defined and neither delayed nor aggressively foisted upon unsuspecting, grieving, and stunned parents.

Care for patients with spinal muscular atrophy is often best accomplished with a multispecialty team approach, when possible. Successful teams have a point person who is mindful of the many needs and can obtain appropriate medical, social, and spiritual assistance as appropriate. In addition, hospice referral or other provision for the specific issues regarding terminal care, grief, and bereavement support is important. In the circumstance of a choice against mechanical ventilatory support, appropriate provision for management of terminal dyspnea can be of comfort to the patient and family alike. Use of nebulized narcotics can avoid much of the concern that overdosing contributes to death and provide comfort to the patient.

Acknowledgments

We thank Dr Edward Giannini for his critical suggestions on the construction of the Delphi questions and surveys. We also thank all SMA experts who participated in the Delphi survey.

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Presented at the Neurobiology of Disease in Children: Symposium on Spinal Muscular Atrophy, in conjunction with the 35th annual meeting of the Child Neurology Society, Pittsburgh, Pa, October 18-21, 2006. This project is supported by grants from the Patient Advocacy Groups of the International Coordinating Committee for Spinal Muscular Atrophy clinical trials. The patient advocacy groups include the Muscular Dystrophy Association, USA (www.mdausa.org); the Spinal Muscular Atrophy Foundation (www.smafoundation.org); FightSMA (www.fightsma.org); Families of Spinal Muscular Atrophy (www.fsma.org); and The Jennifer Trust (www.jtsma.org).

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A **Family Guide** to the Consensus Statement for Standard of Care in **Spinal Muscular Atrophy**

This Family Guide to the **Consensus Statement for Standard of Care in Spinal Muscular Atrophy** was prepared by SMA Advocates for families affected by SMA. The full text of the Consensus Statement (22 pages) was published in the August 2007 issue of the *Journal of Child Neurology* and can be found on the journal's website. A link to the document is provided on the last page of this Guide.

What is SMA?

Spinal muscular atrophy (SMA) is a rare, inherited disease that results in loss of nerves in the spinal cord and weakness of the muscles connected with those nerves. The muscles most frequently affected are those of the neck and trunk that control posture, those of the leg and arm that control movement, and those in the area of the ribs that help breathing.

People with SMA generally appear normal at birth; the symptoms develop as early as 3 months in the most severely affected, around 1 to 2 years of age in the moderately affected, and more rarely in late childhood or adult years in mildly affected individuals.

There is no known treatment for SMA; historically, nearly half of babies born with the most severe form of the disease have died before age two. All people with SMA have a higher than normal risk for progressive disability. The most severely affected are at risk for breathing complications and premature death.

What is a Consensus Statement and Why Do We Need One?

A consensus statement reflects general agreement among a group. In this case, the Consensus Statement for Standard of Care in Spinal Muscular Atrophy (SMA) was developed by a group of experts in the care of people with SMA. The goal of the consensus statement is to serve as a resource for healthcare professionals and to provide recommendations for the most current treatments. This is especially important for people living with SMA, as many communities simply do not have access to specialists and experts in SMA care.

The following is a quick guide to the contents of the consensus statement especially for families and patients. We hope it will alert you to topics for discussion with your doctors and healthcare team. *It is very important to understand that these are suggestions — the consensus recommendations are for your general consideration and should not be considered absolute requirements for care.* We hope you will share this guide with SMA families and friends. The full Consensus Statement document (see reference on page 4) is available on the journal's website for you and your care team.

What Do the Experts Recommend for Care of Children with SMA?

It is important to know that there are things you can do to keep your child with SMA comfortable and safe and to help him or her learn and grow to fullest capacity. SMA experts recommend five key areas for discussion with your doctors/therapists:

1. **Confirming the diagnosis of SMA**
2. **Managing breathing**
3. **Managing eating and nutrition**
4. **Managing movement and daily activities**
5. **Preparing for illness**

1 Confirming the Diagnosis of SMA

If you are reading this information sheet, you and your medical team are probably already talking about the possibility that your child is affected by SMA. A simple blood test can confirm whether or not your child has a mutation in the SMN gene that causes SMA. If the gene test is positive, then your child's diagnosis is established. However, about 5% of children with symptoms of SMA can have a negative SMN gene test and may require further diagnostic tests. These tests can include an electromyography study (EMG), a nerve conduction study (NCS) and/or a muscle biopsy and blood tests to help rule out other forms of muscle disease.

If the blood test comes back positive for SMA, your doctor will talk with you and your family about planning for the care of your child, including plans for supporting breathing and feeding as well as physical and occupational therapy. Genetic counseling is also recommended for families affected by SMA.

The chart describes the spectrum of SMA seen in the clinic. It is used as a guide for discussion and planning by SMA experts when they are working with families and the heathcare team. The classification by onset of weakness and functional level can help you and your doctor understand how much your child will be affected by SMA. Generally, early weakness is associated with more severe disease and a greater need for proactive care planning.

The need for individualized planning for all people with SMA cannot be emphasized enough – it will help you and your family understand and prepare for daily life and how to respond to medical emergencies you might encounter in the course of your child's disease.

Clinical Classication of SMA *(commonly used by health professionals)*

SMA Type	*Highest Function Attained*	*Age of Onset (Weakness First Noticed)*
Type I	Never sit independently	0-6 months
Type II	Can sit without support; cannot walk independently	7-18 months
Type III	Can stand and walk independently; may require aids	>18 months
Type IV	Walks during adult years	2nd or 3rd decade

REMEMBER

- If SMA is suspected, ask for a blood test to confirm or rule out the disease
- If SMA is confirmed, talk with your doctor or a genetic counselor about what this means for your family
- Learn more about the specific medical problems associated with SMA and work with your medical team to develop a care plan personalized for you and your child

2 Managing Breathing Problems

Respiratory or breathing problems are the number one cause of illness for people affected by SMA and are the most common cause of death among children with Type I and II disease. The muscle weakness associated with SMA makes it difficult to cough and clear lung secretions and can cause poor breathing during sleep. Muscle weakness can make your child more susceptible to respiratory infections. Repeated infections can permanently damage lung tissue and make breathing even more difficult.

Respiratory care of patients with SMA is essential to their survival and quality of life. It is critical that parents and patients be able to recognize breathing problems early and understand the steps needed to help improve breathing.

Since respiratory care planning is so important, discussions with a pulmonologist (a lung and breathing specialist) familiar with SMA management issues should take place as soon as possible after diagnosis. The doctor will evaluate your child's breathing and ability to cough effectively and make recommendations for helping to keep your child's airway clear.

Depending on disease severity, tools that may help include manual or mechanical cough assist devices and non-invasive ventilatory support such as bi-level positive airway pressure devices (BiPAP). Protocols have been developed for the most effective use of these tools for people with SMA and are available from many SMA clinics. Routine immunizations and flu shots are also recommended.

The pulmonary specialist will work with families to develop respiratory care goals personalized for each child. Specific attention should be given to planning for times when your child is sick with colds or flu or in the event of surgery. Plans should be reviewed and revised periodically as your child grows older or as health status changes over time.

REMEMBER

- Learn about maintaining a clear airway – it is important for all people with SMA
- Discuss breathing support options with your medical team as needed
- Work with the medical team to develop a care plan to **prevent** problems
- Work with the medical team to develop a care plan for use during **acute respiratory illness** such as a cold or flu, or in case your child develops more severe problems requiring hospitalization

3 Managing Feeding Problems

People with SMA can suffer from under- or over-nourishment and both can affect your child's quality of life. Your medical team should regularly monitor your child's growth and work with you to develop an optimal nutritional and feeding plan personalized for your child.

It is important to recognize that swallowing difficulties may increase the risk of inhaling food or drink, which may cause recurrent chest infections (aspiration pneumonia) that can be serious and life-threatening complications for people with SMA. Mealtime strategies can be adopted to help prevent food aspiration. Gastric reflux (food coming back up from the stomach) can also cause aspiration pneumonia and preventive measures should be discussed with your medical team.

For children with Type II and Type III SMA, weight gain can be challenging, resulting in further stress on weak muscles and potentially more disability. SMA children are also often affected by constipation. Many strategies have been developed to help children with these eating or digestion problems. A dietician and therapists specializing in speech, swallowing and physical movement can help the medical team determine the best strategies for you and your child.

REMEMBER 3

- ◊ Monitor your child's growth on a growth chart and work with your medical team to develop a personalized feeding plan
- ◊ If your child experiences trouble swallowing, has gastric reflux or is bothered by constipation, talk with your team about developing workable solutions
- ◊ Monitor for eating or digestive problems and develop strategies for preventing aspiration pneumonia

4 Managing Movement and Daily Activities

Muscle weakness, the most obvious symptom of SMA, varies from person to person, depending on disease severity. Care plans for managing muscle weakness of the arms, legs, trunk and neck are important for helping your child achieve his or her highest level of function and independence.

Physical therapists, occupational therapists, speech therapists and/or rehabilitation specialists are the experts who can help you and the rest of the medical team design the best plan for your child. Their evaluations may include range-of-motion, strength and mobility tests. They can recommend exercises and tools or assistive devices to help your child maintain the best posture for lung function and eating, as well as tools for moving during work or play. These tools can also help manage and prevent muscle contractures, spinal deformity, pain and bone fractures that can make your child's disability worse.

Assistive devices ranging from orthotics and braces to motorized wheelchairs are important for your child's health and daily activities. Consulting with a therapist who has experience in working with SMA patients can be helpful in determining which assistive devices are most appropriate for your child and how best to incorporate them into daily living. Children with SMA should be evaluated by an orthopedic doctor for spinal curvature; surgical stabilization of the spine may be recommended.

REMEMBER 4

- ◊ Work to develop a physical therapy plan to help your child achieve their highest personal level of function and independence
- ◊ Consider use of assistive devices, tools and exercise to **support** breathing, eating, work and play
- ◊ Consider use of assistive devices to help slow or prevent complications of SMA
- ◊ Consult with your medical team about evaluation by an orthopedic specialist

5 Preparing for Illness

Caring for your children is challenging enough in normal circumstances; the complications of SMA can add another layer of complexity to family life. In discussing care plans for your child, you and your medical team are likely to talk about daily or chronic care, prevention and what to do in case of medical emergencies. In many cases, *planning* is the single most important thing you can do to prevent a medical emergency. When a medical emergency occurs, the support of a knowledgeable care team is essential. Whether done in advance or in a time of crisis, the most important thing to do is talk with the medical team about care options and their consequences for your child and your family.

Given the lack of specific treatments for SMA, most care or therapy choices for SMA (like the ones discussed in the consensus statement) are considered "supportive interventions." When delivered in a planned manner, these interventions are designed to help your child lead the fullest life possible. In the most severe cases of SMA, however, parents can be faced with anguishing decisions regarding therapies that may be perceived as prolonging suffering rather than relieving the burden of disease.

Whenever possible, end-of-life care options need to be defined and discussed openly with you and your family so that any decisions made reflect your values. It is important to have this discussion, however difficult, before a crisis occurs, so that the medical team is prepared and can work through a medical emergency together with you and your family. You may also want to inform emergency room staff at your local hospital of your plans.

REMEMBER 5

- ◊ Develop a plan for medical emergencies
- ◊ Share your plans and management goals with all the healthcare professionals involved in caring for your child
- ◊ Maintain a notebook or folder of current treatment plans and your decisions about critical care, to help you and healthcare professionals during a medical emergency

The Experts Agree ...

Recommendations, Not Rules

SMA experts agree: Care for children with SMA is often best accomplished with the help of many specialists and primary care providers. Parents are key members of this team and are encouraged to participate as much as possible. The recommendations made in this Family Guide are based on the guidelines developed for doctors and healthcare specialists, but they are only suggestions. **You and your medical team are the best people to decide what is appropriate for your child with SMA. Please contact your doctor with any questions you might have about the consensus guidelines or care for your child.**

TO ACCESS THE ORIGINAL PAPER

Consensus Statement for Standard of Care in Spinal Muscular Atrophy

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Journal of Child Neurology, Volume 22 Number 8, August 2007, 1027-1049.

Consensus statement available online: http://jcn.sagepub.com/cgi/reprint/22/8/1027

Journal of Child Neurology

ICC
International Coordinating Committee for SMA Clinical Trials

This document was prepared by the Patient Advisory Group of the International Coordinating Committee for SMA Clinical Trials

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UW Health

Health Facts *for you*

Cough Assist Machine

The Cough Assist Machine helps to clear secretions from the lungs by helping you with your breathing. When you breathe in, the machine gives you air to help expand your lungs. When you blow out, the machine creates a sucking force that pulls air out of your lungs. This rapid change in pressure during breathing helps make your cough stronger and better.

Machine Set-up

We will help you to set up the machine and learn to make changes on it if needed. You will start with low pressures and then build up to the highest amount of pressure that you can handle during each phase of your breathing. At first, we will set the time for each phase of breathing to your comfort level.

How do I adjust the phases of breathing?

Each phase of breathing should be set so that you can get the air in and out of your lungs with comfort, and then pause to keep coughing if needed.

1. Set the inhale time knob at 1 second.
2. Set the exhale time knob at 1 second.
3. Set the pause time knob at 2 second.
4. Set the inhale flow knob at the 3 squiggly arrows (Full flow)

How do I use the Cough Assist Machine?

1. Check to make sure that the inspiratory (positive) and the expiratory (negative) pressures are set. Start at +20/-20 and gradually increase to +40/-40.
   - Remove the face mask or mouthpiece and hold the tubing firmly against a tight surface.
   - Turn on the machine.
   - Set the manual/auto switch to **manual**.
   - Toggle the manual control switch between inhale and exhale a few times to ensure that the pressures are correct and that the manometer returns to zero.

2. Set the manual/auto switch to **auto**. The unit will then cycle from the positive to negative pressure and back to zero for the pause on its own.

3. Put the mask or mouthpiece back on to the tubing and place it tightly on your face or mouth so that no air leaks out. You may need to use a nose clip to prevent air leakage.

4. Turn the machine on.

5. As you breathe in, the machine will help by giving a big breath of air. This breath will end at the time set on your machine.

6. As you breathe out, the machine will help by sucking the air out. This will help your cough be much stronger and it will help to remove phlegm.

7. Keep taking a series of 4-6 breaths from the cough machine and then rest for 20-30 seconds and cough out any phlegm that may have loosened up.

8. Repeat the series of breaths with a rest 4-6 times or until you no longer have any phlegm to cough out.

As you get used to the pressures during breathing, you will want to increase the pressure to the highest amount that you can handle.

How do I change the pressures?

To get the highest amount of pressure you can tolerate, you must change the pressures on the cough machine.

1. Remove the face mask or mouthpiece and hold the tubing firmly against a tight surface.
2. Turn on the machine.
3. Set the manual/auto switch to **manual**.
4. To change both inspiratory pressure and expiratory pressure, turn the Pressure knob clockwise to increase the pressures and counterclockwise to decrease the pressures.
5. To adjust the INSPIRATORY PRESSURE to be less than the expiratory pressure:
    - Toggle and hold the manual control lever to inhale.
    - Decrease INSPIRATORY PRESSURE by turning the Inhale Pressure knob counterclockwise to the positive pressure on the manometer that you can handle.

Once you get to the highest amount of pressure that you can handle, you will no longer have to adjust the pressures. If you need to lower the pressures at any time, you should tell your doctor.

How do I adjust the flow?

If you feel there is too much air flow coming from the machine when you take a breath in, you can decrease the inhale flow.

1. Set the inhale flow knob at one squiggly arrow (reduced flow)

How do I clean my equipment?

To Clean the Mouthpiece

The mouthpiece must be **cleaned after every treatment**.

1. Wash your hands.
2. Run hot tap water for 2 minutes to ensure clear water from tap.
3. Take off the mouthpiece and wash in dish soap (Ivory®, Joy®, etc.) and water in a clean container. Do not wash with your dishes.
4. Rinse with hot tap water
5. Air dry on a clean towel until your next treatment.

Things to know about the Cough Assist Machine

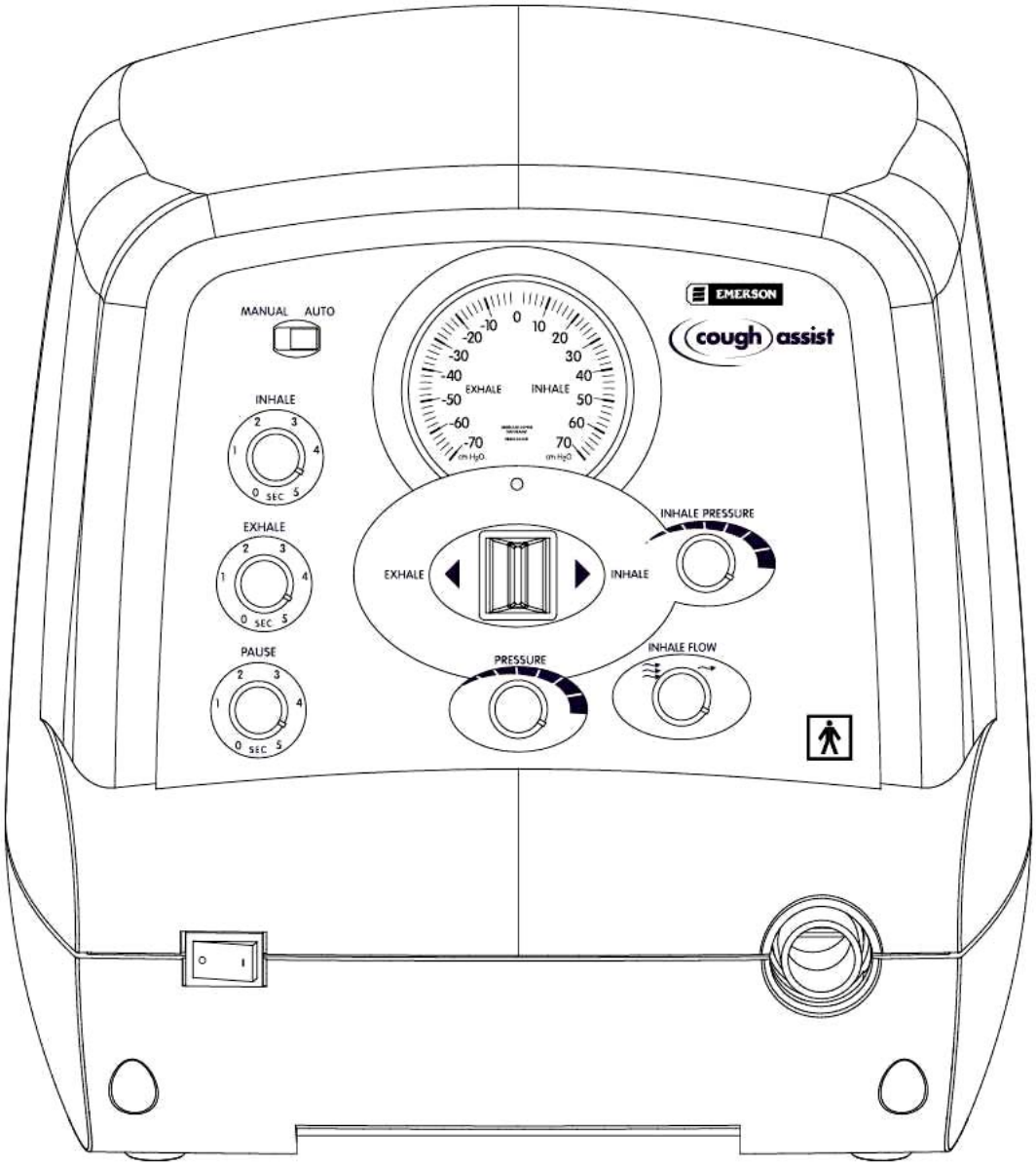
The machine is very easy to learn once you know what each knob controls.

- **Power Switch:** 'I' symbol means ON. 'O' symbol means OFF
- **Inhale**: sets how long the positive pressure will be blowing air into the lungs.
- **Exhale**: sets how long the negative pressure will be sucking or forcing the air out of the lungs.
- **Pause**: sets the time between the last negative pressure and next positive pressure.
- **Inhale Flow**: where you set the amount of flow needed during breathing in. There are two settings: FULL or REDUCED. If REDUCED flow is used, there is less pressure.
- **Inhale Pressure**: where you can adjust the inhale pressure to be less than the exhale pressure.
- **Manual/Auto**: controls whether you want the machine to switch between pressures on its own or you want to do it by hand.
- **Manual Control**: allows you to toggle between inhale and exhale when you are using the manual setting.
- This machine is designed for short term use only. It should not be on straight for more than 5 minutes.

If you have questions about your treatment, please call Regional Services at 1-888-663-7043.

If you have questions about the machine, contact your equipment provider.

The Spanish version of this HFFY is 6366.


MANUAL AUTO
INHALE
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-10 0 10 20 30 40 50 60 70
-20 -30 -40 -50 -60
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cough assist
INHALE PRESSURE
PRESSURE
INHALE FLOW

Eur Respir J 2003; 21: 502–508
DOI: 10.1183/09031936.03.00048102

European Respiratory Journal
ISSN 0903-1936

Cough augmentation with mechanical insufflation/exsufflation in patients with neuromuscular weakness

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Cough augmentation with mechanical insufflation/exsufflation in patients with neuromuscular weakness. M. Chatwin, E. Ross, N. Hart, A.H. Nickol, M.I. Polkey, A.K. Simonds. ©ERS Journals Ltd 2003.

ABSTRACT: Adults and children with neuromuscular disease exhibit weak cough and are susceptible to recurrent chest infections, a major cause of morbidity and mortality. Mechanical insufflation/exsufflation may improve cough efficacy by increasing peak cough flow. It was hypothesised that mechanical insufflation/exsufflation would produce a greater increase in peak cough flow than other modes of cough augmentation. The acceptability of these interventions was also compared.

Twenty-two patients aged 10–56 yrs (median 21 yrs) with neuromuscular disease and 19 age-matched controls were studied. Spirometry was performed and respiratory muscle strength measured. Peak cough flow was recorded during maximal unassisted coughs, followed in random order by coughs assisted by physiotherapy, noninvasive ventilation, insufflation and exsufflation, and exsufflation alone. Subjects rated strength of cough, distress and comfort on a visual analogue scale.

In the neuromuscular disease group, mean±SD forced expiratory volume in one second was 0.8±0.6 $L \cdot s^{-1}$, forced vital capacity 0.9±0.8 L, maximum inspiratory pressure 25±16 cmH_2O, maximum expiratory pressure 26±22 cmH_2O and unassisted peak cough flow 169±90 $L \cdot min^{-1}$. The greatest increase in peak cough flow was observed with mechanical insufflation/exsufflation at 235±111 $L \cdot min^{-1}$ ($p<0.01$). All techniques showed similar patient acceptability.

Mechanical insufflation/exsufflation produces a greater increase in peak cough flow than other standard cough augmentation techniques in adults and children with neuromuscular disease.

Eur Respir J 2003; 21: 502–508.

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Keywords: Noninvasive positive pressure ventilation, paediatrics, physiotherapy, respiratory muscle tests

Received: June 5 2002
Accepted after revision: October 4 2002

M. Chatwin was funded by a grant from the Jennifer Trust for Spinal Muscular Atrophy, Stratford-upon-Avon, UK. Support for patient expenses was provided by Brompton Breathers Trust Fund, London, UK. E. Ross was funded by the Cystic Fibrosis Trust (Bromley, UK), A.H. Nickol by the British Lung Foundation (London, UK) and N. Hart by the Association Française Contre Les Myopathies (Paris, France).

Effective cough is a protective mechanism against respiratory tract infections, which are the commonest cause of hospital admission in patients with respiratory muscle weakness due to neuromuscular disease (NMD) [1]. These patients may exhibit impaired cough and a reduction in peak cough flow (PCF) as a result of inspiratory and expiratory muscle weakness, which causes a reduction in the pressure available to drive the cough manoeuvre. Bulbar dysfunction producing an inability to rapidly open the glottis and maintain patency of the upper airway during cough may decrease the PCF, even in the presence of normal respiratory muscle function. Although, in most patients with NMD, the degree of inspiratory and expiratory muscle dysfunction is similar [2], in some conditions, such as spinal muscular atrophy, expiratory muscle weakness may be the predominant feature [3].

The current recommendation for airway clearance during chest infections in patients with respiratory muscle weakness is intensive physiotherapy. This consists of a modified active cycle of breathing technique accompanied by physical procedures, such as percussion and shaking, and manually assisted cough [4]. However, intensive physiotherapy is tiring for patients and vigorous physiotherapy can precipitate episodic oxygen desaturation [5]. To improve tolerance, physiotherapy can be combined with assistance from noninvasive positive pressure ventilation (NIPPV) [6], but this approach may still be insufficient to clear secretions adequately, resulting in some patients requiring invasive ventilation.

An alternative approach is mechanical insufflation/exsufflation *via* a facial mask. The device used has recently received a Conformité Européenne (CE; European conformity) mark, indicating conformity with European Union safety standards, and become widely available in Europe. A mechanical insufflator/exsufflator uses positive pressure to promote maximal lung inflation followed by an abrupt switch to negative pressure to the upper airway. The rapid change from positive to negative pressure is aimed at simulating the flow changes that occur during a cough, thereby assisting sputum clearance. It was hypothesised that mechanical insufflation/exsufflation would increase PCF in patients with NMD. Thus, the aim of the present study was to quantify the magnitude of this effect with mechanical insufflation/exsufflation compared to other cough augmentation techniques commonly recommended for use by the Sleep and Ventilation Unit, Royal Brompton Hospital, London, UK. The aim was also to compare patient (adults and children) tolerance of mechanical insufflation/exsufflation with that of these other techniques.

For editorial comments see page 385.

Methods

Subjects

The study was approved by the local ethics committee and all subjects and/or their parents gave informed consent. All patients (adults and children) had a history of recurrent chest infections and/or ineffective cough. The aim was to study subjects during a period of clinical stability; therefore, those who had required antibiotic therapy within the preceding month and those with a resting arterial oxygen saturation measured by pulse oximetry (S_{p,O_2}) of <90% and/or end-tidal carbon dioxide tension (P_{ET,CO_2}) of >7 kPa were excluded. Additional exclusion criteria were the presence of severe bulbar dysfunction and a previous history of pneumothorax. Age-matched controls were recruited from staff and their families.

Measurements

Sniff nasal inspiratory pressure (SNIP) [7, 8], maximal static inspiratory ($P_{I,max}$) and expiratory ($P_{E,max}$) mouth pressure [9], and maximal whistle mouth pressure ($P_{mo,W}$) [10] were used as noninvasive indices of inspiratory and expiratory muscle strength. During recording of respiratory muscle strength, subjects were seated comfortably in front of a computer monitor, allowing visual feedback during each manoeuvre. Maximal sniff manoeuvres were performed from functional residual capacity with the subject seated comfortably without a noseclip [7]. Pressures were measured at the nose *via* a pressure line inserted into a bung. Repeated sniffs were performed until no further increase in pressure was seen, with the highest value being recorded as the SNIP [11]. $P_{I,max}$ was measured during a maximal inspiratory manoeuvre from residual volume *via* a mouthpiece with a noseclip *in situ*, performed against a closed shutter [12]. $P_{E,max}$ was recorded using the same equipment during a maximal expiratory manoeuvre from total lung capacity (TLC). $P_{mo,W}$ was measured during a maximal expiration of <500 ms from TLC through a reversed paediatric inhaler whistle (Turbohaler trainer; Astra Pharmaceuticals Ltd, Kings Langley, UK) connected to a flange mouthpiece, without a noseclip [10]. Manoeuvres were repeated at least six times until three readings were within 10% of each other. The highest of these readings was recorded. All signals were measured using a differential pressure transducer (Validyne, CA, USA) and amplified. The signals passed *via* an analogue/digital board to a computer running Labview software (National Instruments, Austin, TX, USA). During testing, subjects received strong verbal encouragement [13]. Forced expiratory volume in one second and forced vital capacity were measured using a hand-held spirometer (2120; Vitalograph Ltd, Maids Moreton, UK) [14]. Resting S_{p,O_2} was recorded using a fingertip probe and portable pulse oximeter (NPB-40; Nellcor, Pleasanton, CA, USA). S_{p,O_2} was taken as the highest recorded value measured during a 3–5-min period with the subjects seated in the upright position. P_{ET,CO_2} was recorded while subjects breathed through a full face mask connected *via* a rigid catheter to a capnograph (PK Morgan, Gillingham, UK), with the baseline reading taken as the P_{ET,CO_2} at the end of 5 min of resting breathing.

The primary outcome measures of PCF and patient comfort were evaluated during each test procedure. PCF was measured by coughing into a tight-fitting full face mask (Mirage Full-face mask; Resmed, Abingdon, UK) connected *via* plastic tubing to a metal tube (41 cm long; 3.5 cm internal diameter). The tube was connected to a Fleisch No. 4 pneumotachograph head (Fleisch, Lausanne, Switzerland) and an electrospirometer (GM Instruments, Kilwinning, UK). The pneumotachograph head was connected *via* ventilator tubing to a mechanical insufflator/exsufflator ("Cough-Assist"; JH Emerson Co., Cambridge, MA, USA). Mask pressure was measured from a side port. A technician secured the mask on to the subject's face to minimise air leak.

The comfort of and distress caused by the intervention together with perceived strength of cough were evaluated using a Visual Analogue Scale (VAS) [15]. Following each intervention, the subjects rated comfort of intervention, distress and strength of cough produced (0: least; 10: most).

Cough interventions

All subjects were studied in their preferred position, usually seated. Initial assessment consisted of at least six maximal unaided coughs followed in random order by the cough intervention techniques; at least six maximal efforts were made during each intervention, with rest periods between them. The cough intervention techniques were: standard physiotherapy-assisted cough [4, 16]; cough after inspiration supported by a noninvasive positive pressure ventilator (BiPAP (bilevel positive airway pressure) from Respironics, Inc., Murrysville, PA, USA, or PV401 from Breas Medical, Moinlycke, Sweden) [17]; exsufflation-assisted cough with delivery of negative pressure initiated manually at the end of inspiration; insufflation (given manually during inspiratory phase); and exsufflation-assisted cough with delivery of the negative pressure immediately preceding the cough effort. Use of pressure-limited or preset ventilators meant that patients were unable to "breath stack", with multiple inspirations prior to coughing, as would be possible with volume preset ventilators. Both insufflatory and exsufflatory pressures were titrated to patient comfort.

Statistical analysis

All values are expressed as mean±SD unless stated otherwise. A p-value of ≤ 0.05 was considered significant. Differences in PCF from baseline using different cough-enhancing techniques were analysed using analysis of variance with analysis of significant differences using Tukey's multiple comparison test. Differences in respiratory muscle strength between patient and control groups were compared using a Mann-Whitney U-test as the data were not normally distributed. Spearman's rank correlation coefficient was used to examine the relationship between PCF and inspiratory and expiratory respiratory muscle strength.

Results

Twenty-two patients (six female) and 19 age-matched controls were studied. Their diagnoses were intermediate spinal muscular atrophy (n=10), Duchenne's muscular dystrophy (n=6), poliomyelitis (n=3) and other congenital muscular dystrophies (n=3).

Seventeen of the 22 patients were using nocturnal NIPPV at the time of the study (mean duration of ventilation 49±28 months). Patients had required between two and six courses of antibiotics during the preceding 12 months for respiratory tract infections and all reported poor cough. Eleven of them had severe scoliosis requiring a spinal jacket (Cobb angle of >40°) and 11 had undergone previous spinal surgery.

Inspiratory and expiratory muscle strength, spirometry, pulse oximetry and P_{ET,CO_2} data are shown in table 1 for the patient group and table 2 for the control group.

For all tests of inspiratory and expiratory muscle function, there was a significant difference between patients and controls

Table 1. – Patient demographics

Patient no.	Age yrs	Diagnosis	SNIP cmH_2O	$P_{mo,W}$ cmH_2O	$P_{I,max}$ cmH_2O	$P_{E,max}$ cmH_2O	FEV1 L	FVC L	FEV1/FVC %	S_{p,O_2} %	P_{ET,CO_2} kPa
Paediatric											
1	10	SMA	38.0	40.0	53.2	41.7	1.5	1.7	88.4	98	5.5
2	11	SMA	27.2	25.7	23.7	30.6	0.4	0.4	100.0	95	5.1
3	12	SMA	35.7	15.0			0.2	0.4	50.0	97	5.9
4	14	DMD	21.0	19.0	19.0	16.7	0.4	0.5	73.6	97	5.7
5	17	DMD	10.0	11.2	14.5	9.7	0.4	0.5	72.9	96	4.9
6	15	DMD	18.0	14.6	13.6	15.4	0.4	0.9	44.0	97	6.1
7	14	CMD	29.8	15.1	11.6	7.8	U	U		95	5.3
8	16	CMD	19.0	31.7	23.2	15.9	0.4	0.3	106.1	94	6.0
Mean±SD	14±2		24.8±9.5	21.5±10.0	22.7±14.3	19.7±12.2	0.5±0.4	0.7±0.5	76.41±23.63	96±1	5.6±0.4
Adult											
9	18	SMA	51.1	18.8	34.6	21.1	1.1	1.2	97.4	97	5.0
10	18	DMD	6.8	6.8	7.5	8.1	0.3	0.3	93.5	94	htvf
11	44	SMA	28.1	55.0	30.8	61.9	2.7	3.3	81.9	97	6.0
12	22	SMA	30.6	37.8	63.0	31.5	1.6	2.0	76.7	97	5.6
13	20	SMA	13.3	11.5	12.6	11.9	U	U		94	6.1
14	28	SMA	30.1	16.9	33.3	17.8	0.9	0.9	98.8	97	5.7
15	35	SMA	63.5	20.9	46.8	23.5	1.3	1.4	97.1	98	5.0
16	22	SMA	14.2	11.0	15.5	14.8	0.3	0.4	62.8	98	4.8
17	25	DMD	6.4	4.6	6.9	5.4	0.3	0.5	58.3	96	7.7
18	30	DMD	5.0	4.4	8.9	4.6	U	U		98	7.1
19	22	CMD	14.6	20.0	18.3	23.2	0.3	0.4	94.4	94	5.0
20	56	Polio	36.6	65.0	36.3	52.7	0.6	0.9	70.3	94	5.2
21	53	Polio	28.0	53.0	17.5	47.8	0.9	1.0	86.4	96	5.6
22	45	Polio	45.0	111.5	40.5	94.8	0.7	0.9	81.3	96	5.2
Mean±SD	31±13		26.7±17.9	31.2±30.5	26.6±16.8	29.9±25.8	0.9±0.7	1.1±0.9	83.3±13.9	96±2	5.7±0.9
All patients											
Mean±SD	25±13		26.0±14.8	27.7±24.6	25.3±15.4	26.5±21.9	0.8±0.6	0.9±0.7	80.7±17.3	96±1	5.6±0.7

SNIP: sniff nasal inspiratory pressure; $P_{mo,W}$: whistle mouth pressure; $P_{I,max}$: maximal static inspiratory mouth pressure; $P_{E,max}$: maximal static expiratory mouth pressure; FEV1: forced expiratory volume in one second; FVC: forced vital capacity; S_{p,O_2}: arterial oxygen saturation measured by pulse oximetry; P_{ET,CO_2}: end-tidal carbon dioxide tension; SMA: spinal muscular atrophy; DMD: Duchenne's muscular dystrophy; CMD: congenital muscular dystrophy; polio: poliomyelitis; U: unrecordable.

($p<0.001$). Compared to controls, expiratory muscle strength was lower in both NMD children (mean differences: $P_{E,max}$ 74 cmH_2O, $P_{mo,W}$ 91 cmH_2O; $p<0.001$) and adults (mean differences: $P_{E,max}$ 100 cmH_2O, $P_{mo,W}$ 128 cmH_2O, $p<0.001$), indicating severe expiratory muscle weakness. There was no difference in inspiratory and expiratory muscle strength measurements between the different patient groups ($p=0.89$).

Resting S_{p,O_2} was significantly lower in the patient group than in the control group ($p<0.005$). There was no significant difference between P_{ET,CO_2} for the patient and control groups ($p=0.06$).

The mask pressure for the patient group was 15±3 cmH_2O during insufflation and -15±9 cmH_2O during exsufflation. For the control group, mask pressure during insufflation was 17±5 cmH_2O and -8±11 cmH_2O during exsufflation.

In the control group, there was a significant positive correlation between PCF and $P_{mo,W}$ ($r=0.71$; $p<0.001$) and between PCF and $P_{E,max}$ ($r=0.67$; $p<0.002$). In the NMD group, there was also a significant positive correlation between PCF and $P_{mo,W}$ ($r=0.69$; $p<0.001$) and between PCF and $P_{E,max}$ ($r=0.55$; $p<0.02$). $P_{E,max}$ and $P_{mo,W}$ were better correlated in the NMD group ($r=0.93$; $p<0.0002$) than in the control group ($r=0.85$; $p<0.0005$).

Representative flow traces for each of the interventions are shown in figure 1. PCFs for the NMD patients for each intervention (mean (95% confidence interval (CI)) were: unassisted cough 169 (129–209) $L\cdot min^{-1}$; physiotherapy-assisted cough 188 (146–229) $L\cdot min^{-1}$; noninvasive ventilator-assisted cough 182 (147–217) $L\cdot min^{-1}$; exsufflation-assisted cough 235 (186–284) $L\cdot min^{-1}$; and insufflation/exsufflation-assisted cough 297 (246–350) $L\cdot min^{-1}$. By comparison, the results for the control group were: unassisted cough 578 (508–648) $L\cdot min^{-1}$; physiotherapy-assisted cough 587 (512–663) $L\cdot min^{-1}$; non-invasive ventilator-assisted cough 565 (495–635) $L\cdot min^{-1}$; exsufflation-assisted cough 633 (570–695) $L\cdot min^{-1}$; and insufflation/exsufflation-assisted cough 629 (565–603) $L\cdot min^{-1}$. Analysis of variance for interventions revealed significant differences in all groups: patient group $p=0.001$; control group $p=0.0001$. There was an increase in PCF from baseline (unassisted cough) with exsufflation ($p<0.001$) and insufflation/exsufflation ($p<0.01$) in the control group. In the combined adult and paediatric patient group, an increase in PCF with insufflation/exsufflation was observed compared to exsufflation alone ($p<0.001$). In addition, in the adult NMD patients, analysis of variance for interventions revealed significant differences ($p=0.0001$). There was a difference from baseline PCF with exsufflation ($p<0.01$), and insufflation/exsufflation ($p<0.001$) (fig. 2). The analysis of variance for interventions in paediatric NMD patients revealed significant differences ($p=0.0001$). A significant increase in PCF during mechanical insufflation/exsufflation ($p<0.001$) was also observed. Analysis of variance for interventions revealed significant differences ($p=0.0005$) in the paediatric control group and an increase in PCF during mechanical insufflation/exsufflation ($p<0.01$) (fig. 2). Analysis of variance for interventions revealed significant differences ($p=0.0001$) for the adult control group. Both patients and controls tolerated all of the interventions well and no adverse events were observed. All subjects, in both the patient and control groups, reported the greatest increase in cough strength compared to unassisted cough during insufflation/exsufflation ($p<0.001$). Analysis of variance for interventions revealed significant differences ($p=0.0001$) in mean scores. The VAS results (mean (95% CI))

Table 2. – Control demographics

Patient no.	Age yrs	SNIP cmH_2O	$P_{mo,W}$ cmH_2O	$P_{I,max}$ cmH_2O	$P_{E,max}$ cmH_2O	FEV_1 L	FVC L	FEV_1/FVC %	S_{p,O_2} %	P_{ET,CO_2} kPa
Paediatric										
1	11	65.4	58.3	59.7	31.0	1.4	1.8	78.4	96	5.7
2	10	90.3	100.6	89.0	112.0	1.5	2.0	73.0	96	5.1
3	14	90.0	132.0	90.6	74.7	5.1	6.0	83.9	97	5.3
4	12	105.6	102.5	77.7	78.0	2.6	3.3	78.5	98	5.0
5	14	103.2	70.0	91.3	90.1	2.1	2.8	73.0	98	4.6
6	16	79.0	105.0	125.4	79.1	3.4	3.6	93.3	97	5.1
7	15	104.0	167.7	164.8	123.8	4.5	6.2	72.2	98	5.3
8	17	129.0	170.0	120.0	164.2	2.3	2.7	86.8	97	6.1
Mean±SD	13.6±2.4	95.8±19.2	113.3±41.0	102.3±33.0	94.1±39.6	2.8±1.3	3.5±1.7	79.9±7.6	97±1	5.3±0.4
Adult										
9	18	108.0	136.0	120.5	136.0	2.1	2.5	85.7	98	5.3
10	25	101.0	154.2	90.9	120.6	3.8	4.5	84.0	98	
11	44	116.0	151.0	98.6	108.6	3.0	4.7	65.4	96	5.2
12	22	130.0	208.0	149.8	157.2	4.9	6.0	69.8	98	
13	22	67.0	95.0	48.3	85.5	3.3	3.8	88.3	99	4.8
14	20	118.6	256.0	116.0	181.0	4.2	5.9	70.9	98	5.7
15	18	95.0	114.0	86.1	83.9	3.4	3.7	91.8	99	4.5
16	30	114.5	152.6	161.7	177.5	3.8	4.0	77.3	98	4.0
17	28	43.8	147.2	112.6	125.3	3.5	4.2	83.2	98	
18	35	56.1	78.5	72.1	64.2	3.4	4.3	79.1	97	5.8
19	22	65.2	104.5	81.8	92.4	3.2	3.4	93.6	98	4.5
Mean±SD	26±8	92.3±29.2	145.2±51.1	103.5±33.2	121.1±39.0	3.5±0.7	4.4±1.2	80.8±9.2	98±1	5.1±0.5
All controls										
Mean±SD	21±9	93.8±24.9	131.7±48.6	103.0±32.2	109.7±40.5	3.2±1.0	4.0±1.5	80.4±8.4	98±1	5.2±0.5

SNIP: sniff nasal inspiratory pressure; $P_{mo,W}$: whistle mouth pressure; $P_{I,max}$: maximal static inspiratory mouth pressure; $P_{E,max}$: maximal static expiratory mouth pressure; FEV_1: forced expiratory volume in one second; FVC: forced vital capacity; S_{p,O_2}: arterial oxygen saturation measured by pulse oximetry; P_{ET,CO_2}: end-tidal carbon dioxide tension.

from the NMD patients were: unassisted cough 5.4 (4.5–6.3) cm; physiotherapy-assisted cough 5.9 (5.2–6.7) cm; noninvasive ventilator-assisted cough 5.8 (4.8–6.8) cm; exsufflation-assisted cough 6.9 (5.3–7.0) cm; and insufflation/exsufflation-assisted cough 7.3 (6.6–8.0) cm. Control subjects' VAS results were: unassisted cough 7.0 (6.4–7.7) cm; physiotherapy-assisted cough 7.7 (7.1–8.3) cm; noninvasive ventilator-assisted cough 7.2 (6.5–7.9) cm; exsufflation-assisted cough 7.9 (7.3–8.5) cm; and insufflation/exsufflation-assisted cough 8.1 (7.5–8.6) cm. There was no significant change from baseline in results for comfort or distress of intervention on the VAS.

Discussion

The main finding of the present study is that, in patients (adults and children) with severe respiratory muscle weakness due to NMD, the combination of insufflation and exsufflation produces a higher PCF than a voluntary unassisted cough or a cough assisted by noninvasive ventilation. Furthermore, mechanical insufflation/exsufflation was tolerated as well as the other techniques of cough enhancement.

Limitations of the study

The patient group was highly selected in that it showed evidence of extreme muscle weakness, to the extent that the majority (77%) of the group used domiciliary NIPPV. Thus, the present data cannot be used to support the use of mechanical insufflation/exsufflation in patients with less advanced disease. However, the observation that mechanical insufflation/exsufflation confers an advantage over NIPPV-assisted or physiotherapy-assisted cough is particularly impressive given that the majority of the present patients were experienced ventilator users. In addition, patients with severe bulbar dysfunction were excluded, primarily because of the difficulty in interpreting reduced PCF in this group.

The patients were clinically stable and different results might be obtained in patients who are acutely unwell. Previous work suggests that respiratory tract infections can cause a reduction in expiratory muscle strength [18], in which case mechanical insufflation/exsufflation and/or the other techniques applied might confer even greater benefit. In addition, NIPPV-assisted physiotherapy may be helpful in patients with respiratory failure as a consequence of chest infection.

When mechanical insufflation/exsufflation is used in clinical practice, it has been stated that pressures of -40–40 cmH_2O are necessary for clinical efficacy [19, 20]. The pressures delivered *via* the insufflator/exsufflator in the present study, measured at the facemask, were lower than this. "Cough assist" was used in line with the manufacturer's recommendations in a study group who were unfamiliar with this device. However, in spite of the relatively modest pressures applied in the present study, it was still possible to demonstrate a significant increase in peak cough flow. This study was designed to achieve a physiologically significant result, rather than to study the clinical effects of varying forms of cough augmentation. Identification of the optimal clinical pressures required to enhance cough is a subject for future prospective clinical study.

Barometric (pressure preset) ventilators were used in preference to volume preset ventilators, which may increase augmentation of tidal volume [21], because it was felt preferable to use a ventilator that the individual was familiar with, and pressure preset models are the machines most widely available in the UK.

The position in which physiotherapy was performed was not standardised. It was ensured that patients were studied in the most comfortable position; this meant only two were supine. The authors simulated their own clinical practice in that airway

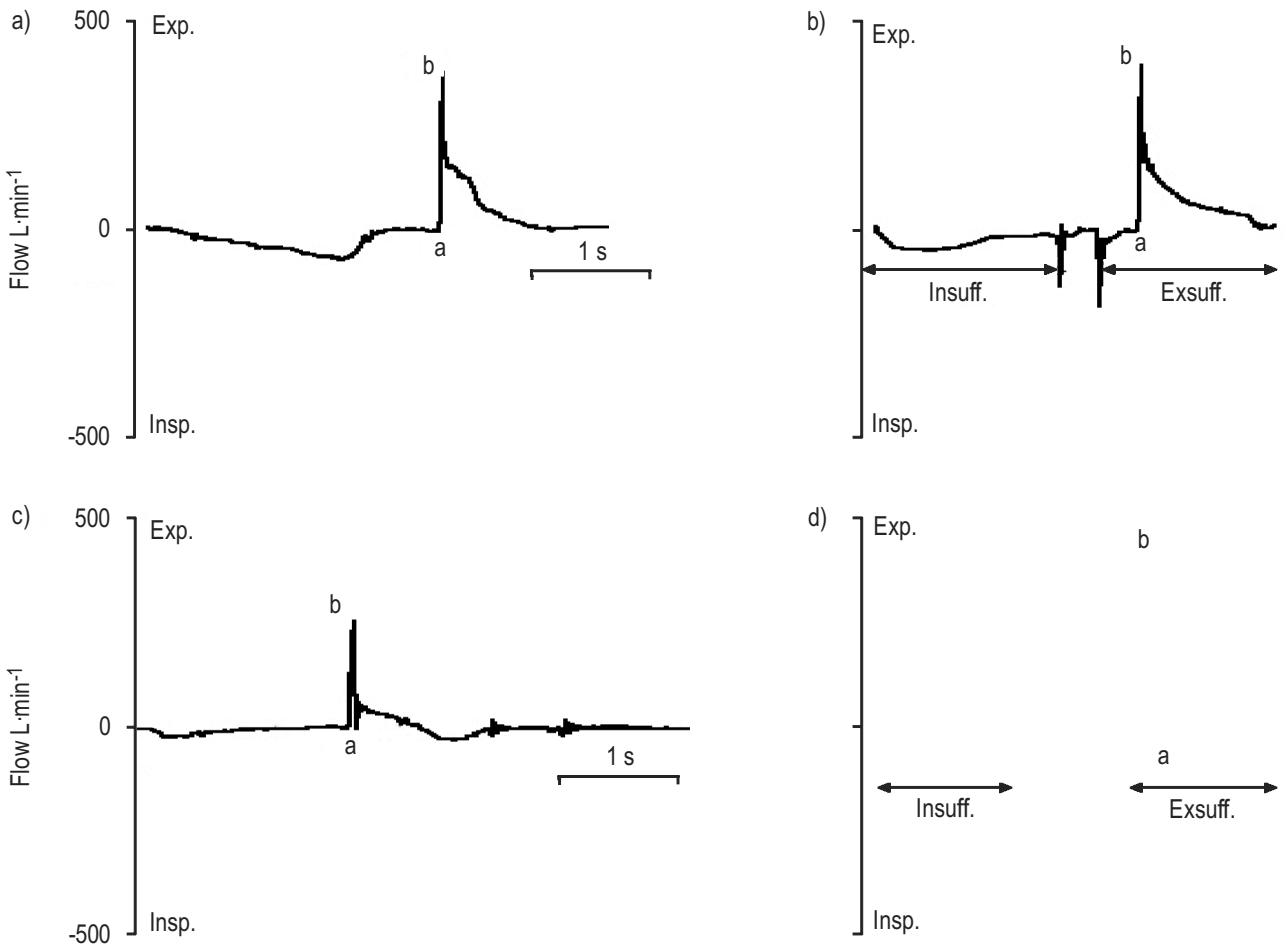

Fig. 1. – a, c) Maximal unassisted cough and b, d) insufflation/exsufflation cough in a, b) a control and c, d) a child with neuromuscular disease. a) represents the start of the cough and b) peak cough flow. The two downward spikes are flow artefact. Note the poor inspiration by the paediatric patient (c). Exp.: expiration; Insp.: inspiration; Insuff.: insufflation; Exsuff.: exsufflation.

clearance techniques were performed in the patient's preferred body position, which encouraged maximal patient cooperation. However, it is acknowledged both that posture influences expiratory flow mechanics directly [22] and some techniques, for example manually assisted cough, may be more effectively applied in the supine position.

Significance of the findings

It is of interest that $P_{mo,W}$ correlated more closely with PCF than $P_{E,max}$. This observation is consistent with a previous study in adults with amyotrophic lateral sclerosis in which $P_{E,max}$ was a poor predictor of the ability to generate transient supramaximal flow [2]. It has been hypothesised previously that $P_{E,max}$ may be a poor screening tool as patients find it hard to generate an airtight seal around the mouthpiece. Observations from the current study suggest that subjects found $P_{mo,W}$ manoeuvres easier to perform. No correlation was found between SNIP and P_{ET,CO_2} but this may be due to the fact that most of the present patients were receiving long-term nocturnal NIPPV, which might influence daytime arterial carbon dioxide tension.

A cough gastric pressure threshold (50 cmH_2O), which it is necessary to exceed to generate transient supramaximal flow, has previously been identified [22]. Although cough gastric pressure was not directly measured in the present study, a relationship between cough gastric pressure and $P_{mo,W}$ has been recently demonstrated [10], and, on this basis, it would be expected that all but two of the present patients would have been unable to produce transient supramaximal flow during an unassisted cough. It is acknowledged that the best cough assist technique for the individual may be dependent on the degree and distribution of respiratory muscle weakness, as well as other factors, including the presence of irreversible expiratory airflow obstruction.

With mechanical insufflation/exsufflation, a large increase in mean PCF was noted in the NMD group (76%). A smaller but still significant increase of 9% was observed in the control group. Given that the control subjects were not flow-limited, the observed increase in PCF is in keeping with the expected increase in transpulmonary pressure caused by mechanical insufflation/exsufflation. However, it should be stated that this increase was small in magnitude, in contrast to that in patients with NMD, and does not suggest a clinical role for mechanical insufflation/exsufflation in patients with normal expiratory muscle strength. The value of insufflation/exsufflation in obstructive lung disease was not assessed in the present study. Previous work has assessed the changes induced by cough augmentation in adults [19, 20, 23–25]; however, few previous studies have carried out detailed physiological assessment of the effect of cough augmentation in both adults and children with the degree of respiratory muscle weakness observed in the present patient group.

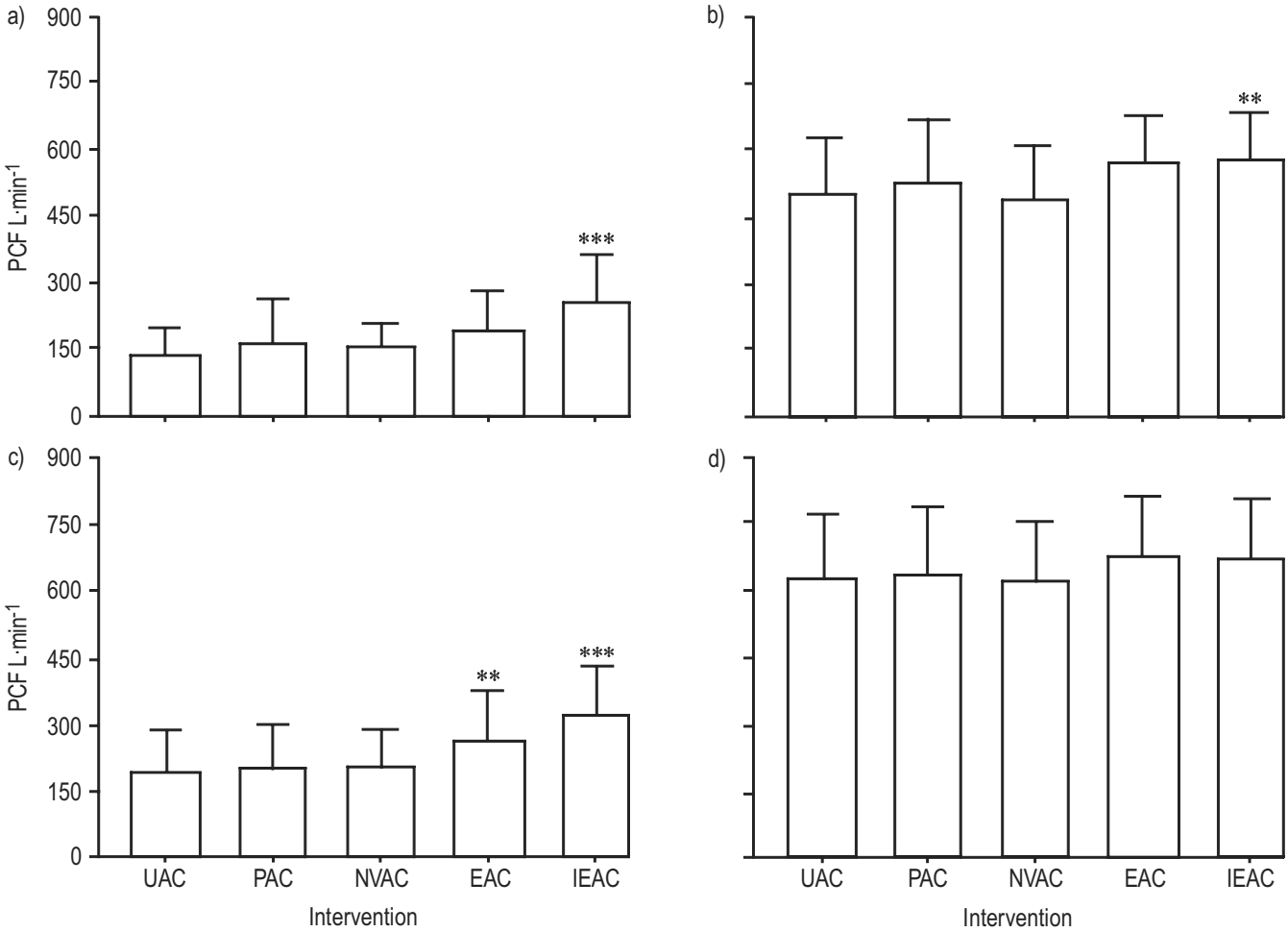

Fig. 2. – Peak cough flow (PCF) in a) paediatric patients, b) paediatric controls, c) adult patients, and d) adult controls. Data are presented as mean± SD. UAC: unassisted cough; PAC: physiotherapy-assisted cough; NVAC: noninvasive ventilator-assisted cough; EAC: exsufflation-assisted cough; IEAC: insufflation/exsufflation-assisted cough. **, ***: $p<0.01$, $p<0.001$ *versus* UAC.

Cough is crucially important for adequate airway clearance. The present study examined the effect on PCF of a variety of interventions and showed that a combination of insufflation and exsufflation causes the greatest increase in PCF. An increase in inspiratory tidal volume is also desirable in mobilisation of secretions, as this enables the generation of higher peak expiratory flows. Patients with respiratory muscle weakness are often unable to generate the increased tidal volume required and repeated attempts to do so may fatigue the individual. To prevent exhaustion, the use of noninvasive ventilation, particularly during acute hypercapnic exacerbations, can provide an increase in tidal volume and therefore aid mobilisation of secretions. However, in the current study, this approach was less effective than mechanical insufflation/exsufflation in increasing PCF.

SIVASOTHY *et al.* [25] studied the effects of cough augmentation, including mechanical insufflation, in subjects with respiratory muscle weakness. The present work differs from their study in several respects. In their study, mechanical insufflation/exsufflation pressures were set at 20 and -20 cmH_2O, two insufflator/exsufflator cycles were used to aid inflation and deflation of the thorax and, after a third inspiration, the subject was asked to make a maximal voluntary cough without the aid of negative pressure. The coughs were, therefore, performed without the aid of exsufflation. The present authors found greater improvement in PCF with the combined insufflation/exsufflation-assisted cough than with insufflation alone. SIVASOTHY *et al.* [25] found that patients with scoliosis showed no increase in PCF and hypothesised that higher insufflation pressures may be required in this subgroup. In the present study, it was ensured that the pressure was titrated to patient comfort and, with this approach, it was possible to show significant benefit in patients with scoliosis. Like SIVASOTHY *et al.* [25], the present authors believe a pneumotachograph gives a more accurate measure of PCF than a standard peak flow meter.

A further notable observation in the present study was that, during titration of mechanical insufflation/exsufflation pressures to patient comfort, the highest insufflation and exsufflation pressures did not necessarily produce the greatest PCF. One reason for this discrepancy could be vocal cord dysfunction, either due to the disease (despite exclusion of patients with moderate or severe bulbar involvement) or as a result of upper airway collapse, secondary to the application of positive or negative pressure during insufflation or exsufflation, respectively [26].

BACH and coworkers [1, 23, 24] have previously suggested that a minimum assisted PCF of 160 $L \cdot min^{-1}$ is required to clear airway debris, where assistance consists of air stacking to maximal insufflation and an abdominal thrust at the time of coughing. Previous retrospective case series [1, 20, 23, 27] from the same group suggest that it may be possible to improve

survival and reduce pulmonary morbidity and hospitalisation rate in patients with a PCF below this level, using noninvasive ventilation and mechanical insufflation/exsufflation, without resorting to tracheostomy. These retrospective data require confirmation in a controlled prospective study.

In conclusion, the present data confirm that a combination of insufflation and exsufflation increases peak cough flow in both children and adults with severe respiratory muscle weakness secondary to childhood-onset neuromuscular disease, in the absence of severe bulbar dysfunction. The use of mechanical insufflation/exsufflation is widespread in North America in subjects with neuromuscular disease. Several case series [1, 20, 23, 27] suggest an important role for mechanical insufflation/exsufflation in the prevention of respiratory morbidity in patients with neuromuscular disease. In the absence of prospective evidence, based upon well-designed randomised studies, there may remain considerable obstacles to the widespread adoption of this device within the UK and Europe. Therefore, randomised trials are now warranted, not only to assess whether regular use of an insufflator/exsufflator can reduce the long-term impact of chest infections in these patients but also to investigate whether the combination of insufflation and exsufflation during acute respiratory tract infections can improve clinical outcome more effectively than existing cough augmentation techniques.

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RESPIRATORY CARE CHOICES: ISSUES FOR SMA TYPE I

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Objectives

- Understand how SMA affects breathing
- Learn techniques to help your child’s breathing
- Realize that you have choices in your child’s respiratory care

CHOICES

- Just as you chose to come to this meeting and this session, you have choices in caring for your child.

Respiratory Care

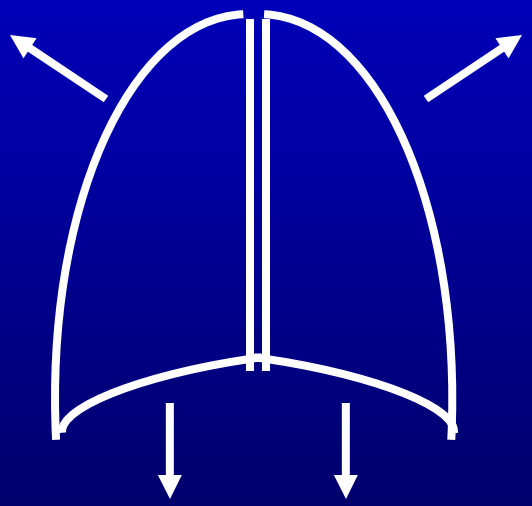
- Critically important!

Respiratory Function in SMA

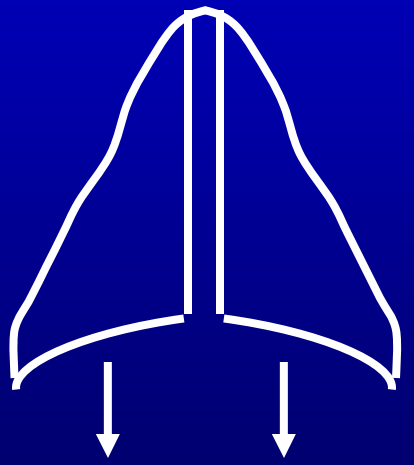
- **Features:**
  - **Very weak intercostal muscles**
  - **Chest wall: very soft and flexible during the first year of life**
  - **Diaphragm: primary muscle used to breath**

Chest Wall Changes

Normal



SMA



Respiratory Muscle Weakness Consequences

1. Lung underdevelopment
2. Difficulty coughing and removing secretions from the lungs
3. Hypoventilation:

   Not taking deep enough breaths to exchange CO2 for oxygen.

Interventions for Respiratory Muscle Weakness Consequences

1. Lung underdevelopment

   Lung expansion daily

Interventions for Respiratory Muscle Weakness Consequences

2. Difficulty coughing and removing secretions from the lungs

Provide techniques to loosen secretions and improve cough

Interventions for Respiratory Muscle Weakness Consequences

3. Hypoventilation

   Breathing support

Measurements of Lung Function

- Pulmonary function testing
  - Infants
    - Pulse oximetry
    - Sleep Study
  - Older children
    - Spirometry (children > 4 years old)
    - Respiratory muscle forces
    - Lung volumes (children > 4 years old)
    - Assisted peak cough flows
    - Sleep Study

Measurements of Lung Function (cont.)

- **Pulse oximetry**
  - **< 95% while AWAKE suggests mucus plugging.**
  - **– Decreased oximetry may be the first evidence of breathing difficulty.**

  - **< 95% while ASLEEP suggests hypoventilation or mucus plugging.**

The Last Straw for SMA Lung Function

- COLDS
  - result in
    - Increased muscle weakness
    - More airway secretions
    - Greater difficulty breathing

Respiratory Care Options

- The answer is not supplemental oxygen!

Respiratory Care Options

- Lung expansion
- Airway secretion mobilization
- Airway secretion clearance
- Breathing support

Lung Expansion

- Bagged or manual insufflations
  - Resuscitator bag with mask
  - In-exsufflator cough machine or Cough Assist device
    - inhalation time at 1-2 seconds
    - exhalation time at 0
  - Mechanical ventilation

Lung Expansion (cont.)

- Intermittent positive pressure breathing (IPPB)
  - Provides synchronized breath support to a preset inspiratory pressure

Lung Expansion (cont.)

- Incentive spirometry
  - Limited to patient's tidal volume
  - Not useful

Respiratory Care Options

- Lung expansion
- Airway secretion mobilization
- Airway secretion clearance
- Breathing support

Secretion Mobilization

- Manual Chest Physiotherapy or Mechanical Percussion
- Postural Drainage
- Intrapulmonary Percussive Ventilation (IPV)
- The Vest Airway Clearance System

Do this before eating or at least 30 minutes after eating.

Respiratory Care Options

- Lung expansion
- Airway secretion mobilization
- Airway secretion clearance
- Breathing support

Airway Secretion Clearance

- Expiratory muscle aids
  - Manual cough assist
  - In-exsufflator cough machine or Cough Assist device

IN-EXSUFFLATOR COUGH MACHINE

- In-exsufflator cough machine improved peak cough expiratory flow rates
  - Mean peak cough expiratory flow rates of 21 patients with NMD
    - Unassisted 1.81 ± 1.03 L/sec
    - Assisted cough 4.27 ± 1.29 L/sec
    - Exsufflator 7.47 ± 1.02 L/sec
    - (normal is 6-12 L/sec)

Bach J. Chest 1993; 104:1553.

IN-EXSUFFLATOR/ COUGH ASSIST DEVICE

IN-EXSUFFLATOR OR COUGH ASSIST

- **SETTINGS to use by mask, mouth piece, tracheostomy tube or endotracheal tube.**
  - **INSPIRATORY**
    - **+25 to +40 cm H_2O for 1-2 sec.**
  - **EXPIRATORY**
    - **-25 to -40 cm H_2O for 1 sec.**
  - **PAUSE TIME**
    - **1-2 sec.**

IN-EXSUFFLATOR OR COUGH ASSIST (cont.)

- Perform 4-5 sets of 5 breaths with a 1-2 minute rest between sets.
- Ideally use manual cough assist with cough machine.
- Suction upper airway or tracheostomy tube or endotracheal tube after coughing.
- Can use every 10 minutes as needed.

Respiratory Care Options

- Lung expansion
- Airway secretion mobilization
- Airway secretion clearance
- Breathing support

Breathing Support

- Indications
  - Standard:
    - Hypoventilation with oxygen saturation <90% and ETCO2 >45

Breathing Support

- Indications
  - SMA
    - Oxygen saturation <94% while asleep and or elevated carbon dioxide level during sleep
    - Chest wall collapse
    - Use of mechanical ventilation in hospital
    - Pneumonia or atelectasis
    - Poor sleep quality

Breathing Support (cont.)

- Options:
  - Negative pressure ventilation
  - Positive pressure ventilation
    - Non-invasive
    - Invasive

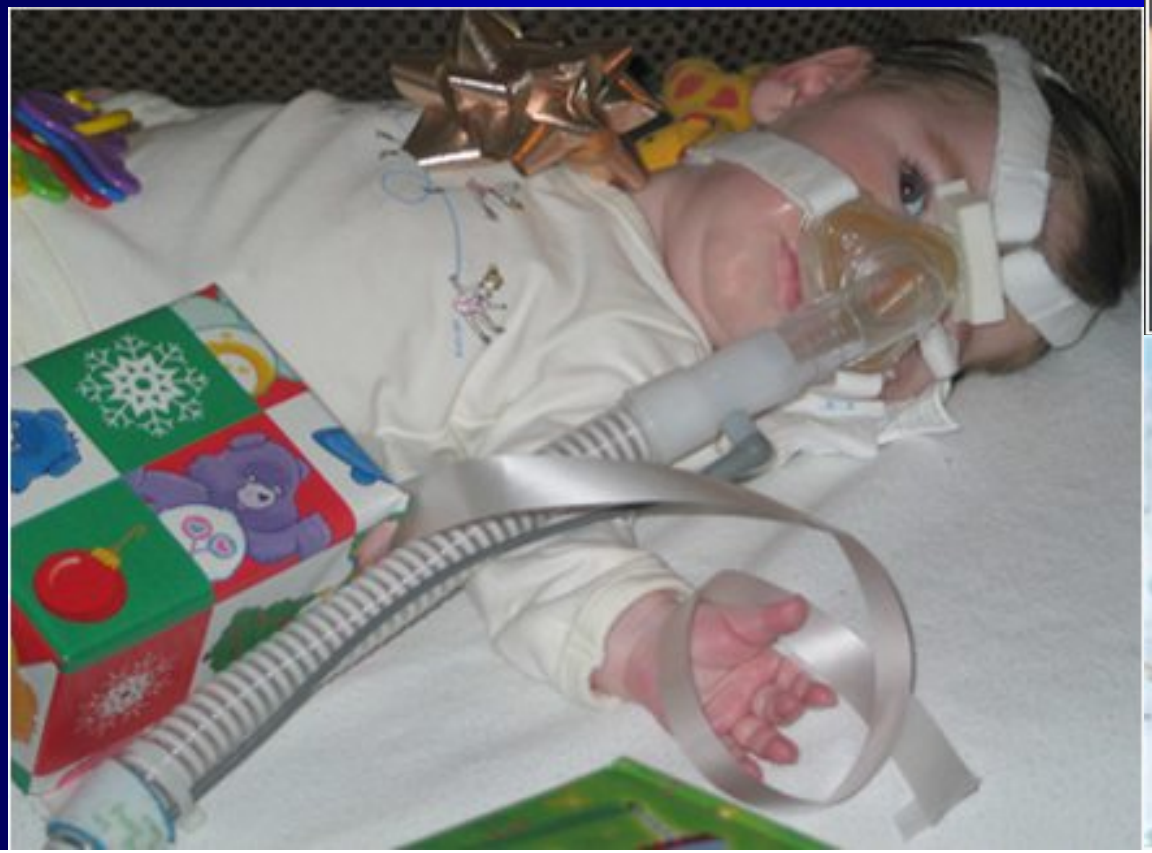
Negative pressure ventilation

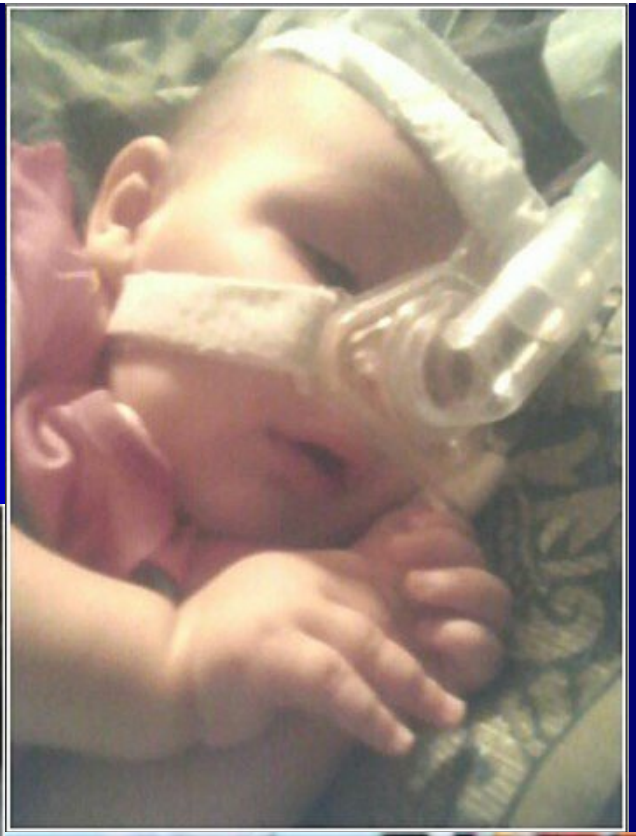
- A device worn over the outside of the chest wall and attached to a vacuum pump
  - Examples:
    - Porta-Lung (Iron lung)
    - Cuirass or Poncho
  - Relative Contraindications
    - Upper airway obstruction

Positive Pressure Ventilation

- Non-invasive
  - Preset air volume or pressure delivered by
    - Nasal mask
    - Full face mask
    - Mouth piece

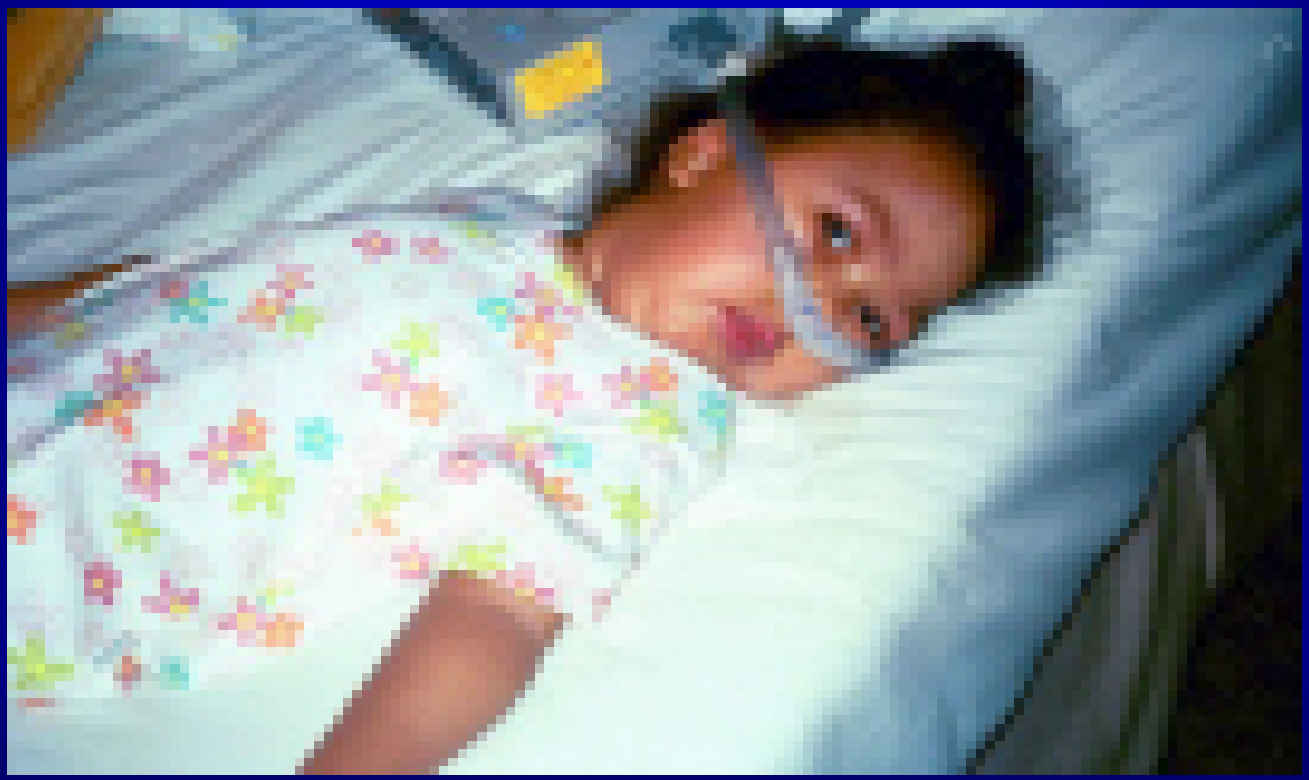
Respironics Small Child Nasal Mask and Head Gear





Respironics Profile Lite Small Child Nasal Mask and Head Gear

Innomed Nasalaire Interface



Non-Invasive Positive Pressure Ventilation

1. Bilevel positive airway pressure (BiPAP).

2. Home mechanical ventilator (LP-6+, LP-10, LTV, Achieva) with volume or pressure ventilation.

Technical Aspects

1. **Bilevel positive airway pressure (BiPAP)**

   Examples: Respironics Synchrony, VPAP III, Knightstar

   IPAP: 14-20 cm of H_20
   EPAP: 3-6 cm of H_20

   Mode: ST (spontaneous timed)

   Respiratory Rate: high enough to capture breathing efforts and rest child.

   Inspiratory Time: depends on pt age and RR

   Rise time: speed of breath delivery

Technical Aspects (cont.)

2. Home mechanical ventilator

Examples: LP-6+,LP-10, LTV 900 or 950
Modes: AC, SIMV PC or SIMV VC

- pressure ventilation
- volume ventilation
- Initial Vt approximately 13-20 ml/kg per nasal mask

NIPPV Disadvantages

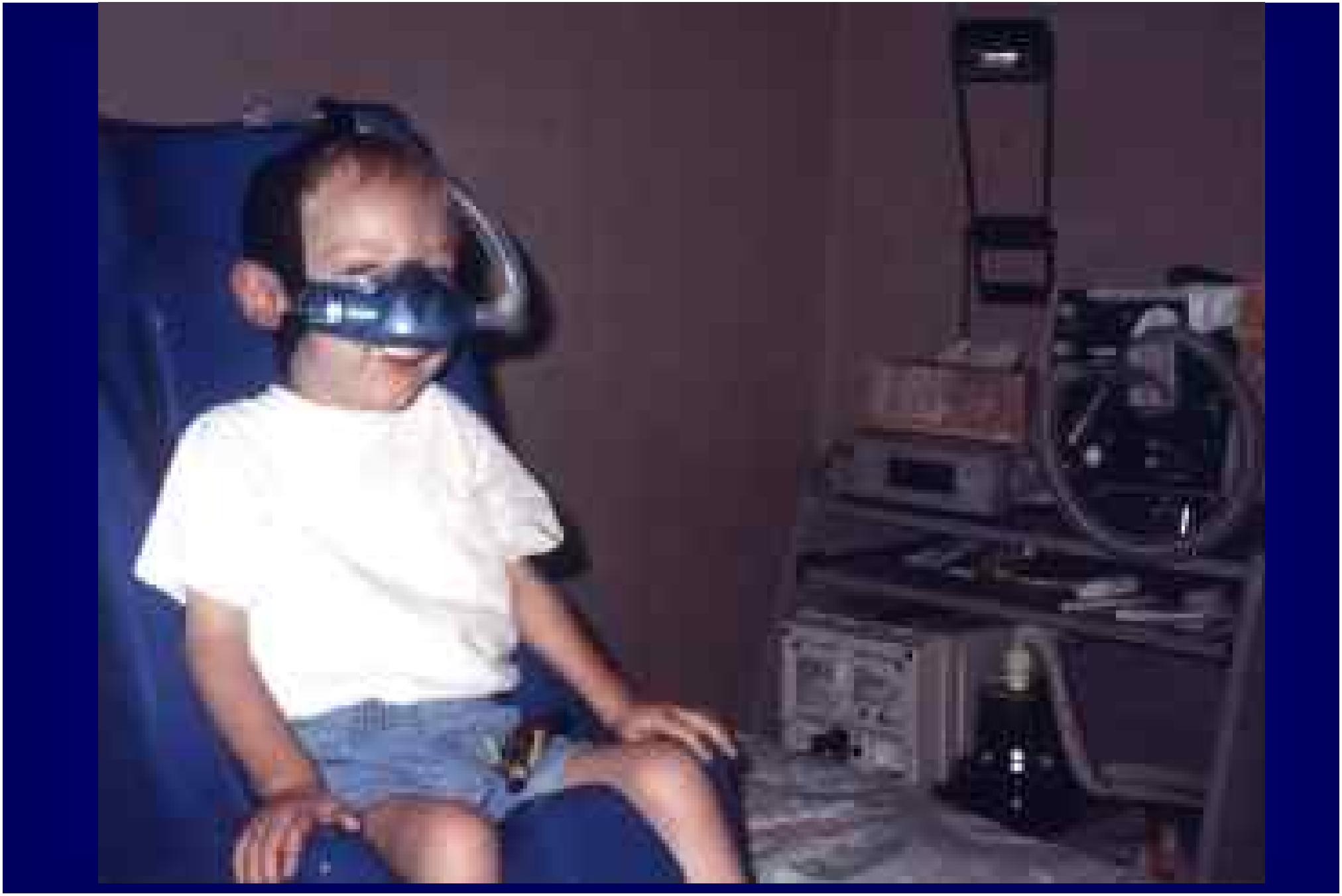
- Gastric distention and emesis especially if children are constipated
- Nasal bridge discomfort and other skin irritation

NIPPV Challenges

- Difficulty swallowing
- Lots of oral secretions
- Gastroesophageal Reflux
- Requiring > 16 hrs/day of ventilation

Recommendations

- **In-home respiratory equipment:**
  - **In-Exsufflator cough machine or Cough Assist**
  - **Suction machine**
  - **Spot check pulse oximeter**
  - **Nocturnal respiratory support**
  - **A consistent method for secretion mobilization, e.g., palm cups, electric percussor, The Vest, IPV**
  - **Postural drainage method.**



Invasive Positive Pressure Ventilation

- Tracheotomy
  - Surgically placed tube through the neck and into the trachea
  - May be necessary for SMA type 1
- Mechanical ventilation

Invasive Positive Pressure Ventilation

- Tracheotomy
  - Reasons
    - Child is not able to tolerate non-invasive ventilation
    - Child requires nose mask ventilation for 24 hours per day when healthy
    - Difficult to manage “death spells”

Care During a Cold

- Every 4 hours
  - Airway clearance for 10-20 minutes
  - In-exsufflator cough machine/Cough Assist
  - Postural drainage for 15-30 minutes
  - In-exsufflator cough machine/Cough Assist

Care During a Cold (cont.)

- Use the In-exsufflator cough machine/Cough Assist as often as needed to clear rattley breathing and help with coughing

Care During a Cold (cont.)

- **Use breathing support during the day and at night.**
  - **REMEMBER YOUR CHILD IS EVEN WEAKER DURING A COLD**

Care During a Cold (cont.)

- Use the pulse oximeter to help guide you in room air.
- When the oxygen saturation is less than 94%, use the cough machine.
- If you cannot keep the oxygen saturation >90%, the child should be seen by a physician.

Care During a Cold (cont.)

- Keep your child hydrated
- Try to continue to feed if possible.

Contributing Factors to Respiratory Muscle Weakness

- Scoliosis
  - Alters chest wall shape and efficiency
- Nutritional status
  - Suboptimal nutrition increases muscle weakness
  - Excess weight contributes to more difficulty moving

Interventions for Contributing Factors to Respiratory Muscle Weakness

- Scoliosis
  - TLSO or Body Jacket
  - Spinal fusion
- Nutritional status
  - Suboptimal nutrition
    - consider gastrostomy tube.
  - Excess weight
    - consider diet limits

CHOICES

Choices

Choices

Choices

Choices

Choices

Families of Children with SMA

- Select community of people who share a common thread . . .

  You all care and love someone with SMA.

CHOICES

Children with SMA type I are very weak in the first year of life and their prognosis is not clear.

RECURRING THEMES

All families struggle with:

- **the diagnosis of SMA**
- **how to best care for their child**
- **whether their child experiences pain as a result of their choices**

CHOICES

- Some children require a breathing tube when they become very ill.

CHOICES

- Some children require a breathing tube when they become very ill.
  - Questions that families and medical care providers think about:
    - Is the condition reversible?
    - Will they come off the respirator/ventilator?

When the Options are Limited

- **Families often have the difficult position of choosing the option they believe is best for their child and their family.**

Because there are no right or wrong choices.

TRUST that you will make the right decision.

Summary

- Children with SMA type 1 are at great risk for poor cough and hypoventilation.
- Many medical interventions are available to support breathing.
- Not all interventions fit every child.

Respironics Profile Lite Small Child Nasal Mask and Head Gear

SleepNet Phantom Nasal Mask and Head Gear