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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