



Caring Choices

For Parents of Infants Newly Diagnosed with SMA Type 1

SMA CARE SERIES



Make today a
breakthrough.

Contents



Dear Parents, Friends and Family Members	3
What Decisions are Ahead	4
What are Care Options?	5
Respiratory	6
GI/Nutrition	9
Palliative Care: What is Palliative Care? -List of things it is/not -Definition of terms	10
Support & Guidance	14
About Cure SMA	15



Dear Parents, Friends and Family Members,

Learning that your child, or the child of someone you care about, has spinal muscular atrophy (SMA) can be a difficult, confusing and worrisome time.

Cure SMA is a not-for-profit organization dedicated to helping families, by offering support, funding research and providing educational resources about this genetic disease.

We hope that this booklet will provide you with useful information and be helpful as you start to think about decisions you will make both today and in the future.

At Cure SMA, we are always here to support you in any way possible.



You can reach us in several ways:

By phone:

1.800.886.1762 (Toll free in the U.S. and Canada)

1.847.367.7620 (all other locations)

By email:

info@curesma.org

On the internet:

We offer a wealth of information for parents and families on the Cure SMA website:

www.cureSMA.org

What Decisions are Ahead?

In the days, weeks, and months ahead, you will be asked to think about many things and to make decisions about your child's care. Some of these decisions may be difficult, but all of them should be made with support, and based on what you feel in your heart is best for your child and your family.

Babies with SMA type I face many physical challenges and often live shortened lives. The medical problems with SMA are caused by muscle weakness which interferes with normal function of the body. In addition to trouble sitting, standing, or walking, the muscle weakness can cause problems in the following areas:

- breathing
- coughing
- swallowing

ALL OF THESE ARE IMPORTANT FOR STAYING HEALTHY

There is no treatment to reverse or stop the progression of SMA type I and at this time, there is no cure. There are, however, care interventions that may help with your baby's muscle weakness and breathing and feeding problems, especially during times of illness.

Together, these interventions can be called "supportive care." These treatments vary in their degree of aggressiveness, and each has risks and benefits.



Each family will make their own "caring choices" when deciding wght for their child.

What are Care Options?

There are multiple treatment options available to support your child. These options range from less invasive to more invasive and every family should make choices that are consistent with their personal beliefs and values and which work best for their child and family. Ask your healthcare team to talk openly with you about what is involved in these different approaches, including the advantages and disadvantages for your child, and what they mean for you, your family, and your child's caregivers.



There is support available from your healthcare team to guide you in making decisions that are best for you, your child, and your family. The options you choose today may change as your child's needs change. Your healthcare team will continue to support you as you may reconsider the treatment options that are right for you and your family. Remember, you can always change your mind regarding the options for your child's care at anytime. You should feel comfortable discussing the various therapies available and your decisions with your healthcare team as questions arise throughout your journey with SMA.



Every child and family with SMA can also benefit from, and should receive, palliative care throughout their illness, regardless of the choices they make. Talk with your healthcare team about what will give your baby the highest quality of life, consistent with your family's beliefs and values. Loving, caring families make different choices and there is no one "right" or best way. This booklet will provide you with an overview of available options and guide you in making decisions that are optimal for your child and your family.



Every child and family with SMA type I can also benefit from, and should receive, palliative care throughout their illness, regardless of the therapies that they choose. Palliative care helps maximize the quality of life for a child living with a serious illness such as SMA.

Respiratory

The early and sometimes most difficult decisions for infants with SMA type I relate to complications with their breathing and nutrition.

Because of the muscle weakness resulting from SMA type I, children will experience early difficulty with their ability to cough and later with breathing itself due to weakness in the muscles that control breathing.

Lung problems pose some of the greatest challenges to infants with SMA type I. Your baby will develop problems with coughing which leads to difficulty clearing their lower airway secretions. As they get older, they will develop difficulty breathing during sleep and ultimately while awake. Due to poor development of the chest wall and lungs, your baby may have difficulty handling the normal colds of childhood. These infections can make already weak muscles for breathing weaker. Children with SMA type I do not develop more respiratory illnesses than children of the same age, but they are more dangerous because of the muscle weakness.

It is very important that your child, you and anyone frequently around your child receive immunizations to help prevent contagious diseases. Infections such as influenza, pneumonia and whooping cough (pertussis) could be devastating for someone with SMA. Fortunately, they are preventable with proper immunizations.

Respiratory care helps with lung function and breathing. With an inadequate cough, children have difficulty clearing secretions which can lead to pneumonia and respiratory failure. A pulmonary/respiratory specialist will work with you to develop respiratory goals that are specific to your child.



WHAT IS NON-INVASIVE RESPIRATORY CARE?

Non-invasive describes a care intervention that does not cut into or go inside the body. Breathing support is placed on or outside the body.

Non-invasive respiratory care will help your child breathe using a mask over the nose. The mask is connected to a “BiPAP” (Bi-level Positive Airway Pressure) machine or a mechanical ventilator.

The breathing support needs of children with SMA type I are variable and hard to predict. Almost all children with SMA type I need help with their breathing and can benefit from a BiPAP machine or ventilator, using a nasal mask, while they sleep, starting at an early age. The BiPAP machine delivers a continuous flow of air to the lungs. During colds, all babies will require additional breathing support. They may need to use their BiPAP or ventilator 24 hours a day for several days until the cold goes away. These non-invasive respiratory care interventions and equipment can be life saving during an illness.



The nose mask does not work well for every child and family, for a variety of reasons, and may become difficult to tolerate. If this is the case, or if your child needs breathing support for 24 hours per day, you will need to make some hard choices about the next steps in your child’s respiratory care plan.



WHAT IS INVASIVE RESPIRATORY CARE?

Invasive means that something goes inside the body as part of the procedure. Needles, tubes, lines and surgery may be needed.

Invasive respiratory care will help your child breathe through a breathing tube attached to a mechanical ventilator. The tube goes into the airways, through either the mouth (referred to as intubation) or through a small hole in the neck (a tracheostomy). A tracheostomy is usually needed if invasive ventilation is required for more than a few weeks.

Initially, invasive respiratory care uses a breathing tube, called an endotracheal tube, which is inserted through the mouth and into the trachea and lungs. Long-term use of endotracheal tubes, however, can cause damage, irritating the mouth and throat if not taken out within a few weeks. In these situations, with ventilator support being needed for more than a few weeks, your child may need to undergo placement of a tracheostomy tube. For this, the doctor creates a small, surgical hole in the neck (a tracheostomy) and inserts a breathing tube (tracheostomy tube) through this hole. This breathing tube, therefore, bypasses the mouth and vocal cords, sparing them long-term damage by going directly into the trachea and lungs.



Supportive Care for Breathing



Less Invasive

More Invasive



BiPAP (or ventilation by mask)

Tracheostomy and ventilator

Please see the book called "Breathing Basics" for more information regarding respiratory care options.

GI/Nutrition

Children with SMA type I usually lose their ability to chew and swallow food and water safely at different ages, depending on the severity of their disease. If they continue to take nutrition and hydration by mouth, they are at high risk for aspirating this food and water and developing respiratory problems (aspiration pneumonia).

To prevent these complications, tube feeding is commonly offered to ensure that babies get enough calories to grow and enough fluids to remain hydrated.

Tube feedings can be given by several routes, including the following:

- nasogastric (NG) tube (a small flexible tube inserted in the nose down to the stomach)
- gastrostomy (G) tube (a tube surgically placed directly into the stomach)

For children with significant gastroesophageal reflux, medication may be used to treat the symptoms, and the following may be recommended to help treat both the inability to eat safely by mouth and reflux:

- g-tube and Nissen fundoplication (stomach wrap)

While the options are invasive, they each have various advantages and disadvantages. NG tubes (or nasoduodenal (ND) or nasojejunal (NJ) tubes) can be easily placed and easily removed, if necessary, but occasionally come out and need to be replaced.

G-tube placement is permanent and easy to maintain, however, does require surgery and anesthesia. The option that is best for your child is determined through discussions with your family and SMA care team.



Supportive Care for Swallowing

← Less Invasive

More Invasive →

Changing Textures of Food
(Temporary tube through nose)

Permanent tube through stomach

Palliative Care

The term palliative care can be frightening or overwhelming for many families as it mistakenly and automatically makes people think only of hospice or end of life (EOL)/death. The information provided here will hopefully help you understand that although palliative care may include options regarding EOL, it is actually not all about death, but rather about living with the best quality of life possible.

The word “palliative” means relieving or soothing symptoms without curing the underlying disease. Palliative care uses an interdisciplinary approach (involves clinicians from different areas) and strives to provide comfort for you and your child and to uphold quality of life, along a physical, psychological, social and spiritual continuum, throughout your child’s life, however long it may be. The philosophy of palliative care ideally should be introduced at the time of diagnosis and provided throughout the illness trajectory as decisions are made on an ongoing basis and as different situations arise.

Having difficult conversations and making well-supported decisions before an emergency occurs will help families be better prepared and avoid making rash decisions at a stressful time. Palliative care prepares families for these situations and should be provided along with whatever treatment options you choose. The palliative care team can help you come up with specific goals that are geared towards what works best for your child and family. Some families may choose invasive measures or non-invasive measures of support, as described above, and some may choose comfort measures only. Some families may choose more or less options than others but all families make choices based on love. Every child and family with SMA can benefit from, and should receive, palliative care throughout their illness, regardless of the choices they make.

The World Health Organization (WHO) defines palliative care as “improving quality of life of patients facing life-threatening illnesses, and their families, through the prevention and relief of suffering by early identification and treatment of pain and other problems, whether physical, psychological, social, or spiritual.”



**FROM THE AMERICAN ACADEMY OF PEDIATRICS,
POLICY STATEMENT ON PALLIATIVE CARE FOR
CHILDREN:**

“Palliative care seeks to enhance quality of life...and the child’s ability to enjoy life... in the face of an ultimately terminal condition.”

“The goal is to add life to the child’s years, not simply years to the child’s life...to optimize the child’s experience, rather than hasten death.”



**FROM THE CONSENSUS STATEMENT FOR THE STANDARD
OF CARE IN SMA:**

“In the most severe cases of SMA, parents can be faced with anguishing decisions regarding therapies that may be perceived as prolonging suffering rather than relieving the...disease.”

“Respect for the dignity of patients and families and preferences regarding testing, monitoring and treatment is a leading principle of palliative care.”



“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.”



PALLIATIVE CARE IS...

- ...making choices that are consistent with your beliefs and values, using your head and your heart.
- ...choosing what is best for your child and your family, and sometimes changing your mind.
- ...a philosophy of care that provides you with support in making difficult decisions.
- ...an interdisciplinary approach to providing your child and your family with the best possible care that meets your goals.
- ...about quality of life, however you and your child define it, from a physical, psychological, social and spiritual perspective.
- ...in addition to treatment, not instead of it.
- ...provides you with the support, information and guidance you need to do what feels best to you and your child.



PALLIATIVE CARE IS NOT...

- ...doing “nothing.”

PALLIATIVE CARE IS...

- ...play.
- ...comfort.
- ...love.
- ...making caring choices.

“You will receive many opinions, some solicited and some volunteered, regarding decisions from medical providers, family members, friends and other families touched by SMA. You may find some of these helpful and others offensive. Please recognize that your choices are just that, yours.

The SMA community is extremely diverse in culture, religious affiliation, political preference and socioeconomic status. Everyone shares the challenges of the disease but the experience of you and your child are unique.”

Palliative Care: Definition of Terms

ADVANCE CARE DIRECTIVES

A set of documents containing instructions that consent to, or refuse, specified medical treatments and that articulate care and lifestyle preferences in anticipating future events or scenarios. This may include “do not resuscitate” (DNR) orders. They become effective in situations where the person is no longer able to make decisions or for children when they are not old enough to make their own decisions. Advance care directives are also, though less frequently, referred to as “living wills.”



ADVANCE CARE PLANNING

The process of preparing for likely scenarios near the end of life that usually includes assessment of, and discussion about, a person’s and family’s understanding of their medical condition and prognosis, values, preferences and personal and family resources. Advance care planning supports patients and families in communicating their wishes about end of life.

HOSPICE

Hospice is a comprehensive service provided to people living with and dying from an eventually fatal condition. This may include inpatient medical care, respite care and end of life care for people who are unable to die at home. Hospices may also offer day care facilities and community home visiting teams.

INTERDISCIPLINARY TEAM

An interdisciplinary team is a team of providers who work together to develop and implement a plan of care. Membership varies depending on the services required to identify and address the expectations and needs of the patient, caregiver and family. An interdisciplinary team typically includes one or more doctors, nurses, social workers, spiritual advisers, pharmacists and personal care workers. Other disciplines may be part of the team if resources permit.

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Where Can You Go for Support and Guidance?

YOUR PRIMARY CARE DOCTOR OR PEDIATRICIAN

Talk first with your primary care provider (PCP) or pediatrician. Even if your PCP is not experienced with the care of patients with SMA, he or she can be an important resource for your family as you decide which “caring choices” you make for your child. Your PCP can contact Cure SMA for help in providing care to you. Ask for referrals to specialists including a pediatric pulmonologist, a pediatric neurologist, a pediatric physiatrist and a pediatric orthopedic surgeon. You may live near a hospital that has a neuromuscular clinic or a clinic specifically for SMA. They will often have all of these specialists located in one place.



THE NEAREST CHILDREN’S HOSPITAL-PEDIATRIC DEPARTMENTS AND SECTIONS

You may want to contact the nearest children’s hospital or the largest medical center in your area and talk with the pediatric neurology department or section.

YOUR CHURCH, SYNAGOGUE, OR MOSQUE, THERAPISTS, FRIENDS

We also encourage you to talk with the spiritual leaders in your place of worship; a psychotherapist, social worker, counselor, or friend—someone who can help you with your feelings and give you a shoulder to lean on. Most hospitals have medical social workers and you can contact them for private conversations.

CURE SMA STAFF, CHAPTERS AND OTHER PARENTS

Talk with us at Cure SMA about finding other parents of children with SMA who have faced these decisions and want to help other parents. We can connect you with families around the country and in your state. We will also support you and your family in any way possible.

CURE SMA WEBSITE

There is the Cure SMA website with so much important information and resources for parents. Visit: www.cureSMA.org.

About Cure SMA

Cure SMA is dedicated to the treatment and cure of spinal muscular atrophy (SMA)—a disease that takes away a person’s ability to walk, eat, or breathe. It is the number one genetic cause of death for infants.

Since 1984, we’ve directed and invested in comprehensive research that has shaped the scientific community’s understanding of SMA. We are currently on the verge of breakthroughs in treatment that will strengthen our children’s bodies, extend life, and lead to a cure.

We have deep expertise in every aspect of SMA—from the day-to-day realities to the nuances of care options—and until we have a cure, we’ll do everything we can to support children and families affected by the disease.

Learn more about how you can help us reach a treatment and cure at www.cureSMA.org.



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We're on the Web!
www.cureSMA.org

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