Musculoskeletal System

SMA CARE SERIES

cure SMA

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Contents

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Introduction:

The goal of this booklet is to help children, families, and other professionals including nurses, physicians, and therapists understand how spinal muscular atrophy (SMA) can affect the body's musculoskeletal system. This booklet is meant to serve as a resource rather than a definitive clinical management guideline. All decisions for the care of a child with SMA need to be made in conjunction with the child, family, and medical or therapy provider and take into account the child's individual medical status and needs.

Throughout the booklet there are recommendations that vary depending on the child's type of SMA. For the purposes of this booklet children are divided into three groups referred to as "non-sitters," "sitters," and "standers/walkers." One could also refer to these groups as SMA "type I," "type II," and "type III" respectively.

INTRODUCTION TO THE MUSCULOSKELETAL SYSTEM:

The musculoskeletal (MSK) system includes the muscles and bones in the body and the supporting structures like tendons (tissue that connects muscle to bone) and ligaments (connects bone to another bone). The MSK system includes the arms, legs, and spine. Weakness associated with SMA can negatively impact the MSK system in the development of the following: limitation in range of motion of the joints, hip dislocation, spinal deformities such as scoliosis, and fractures. Changes in the MSK system such as contractures, fractures, and scoliosis can lead to pain and difficulty with sitting and functional activities. Reducing the risk of MSK problems may be accomplished through use of therapy, orthoses (braces) and equipment.

WHAT ARE BASIC ELEMENTS OF GOOD MUSCULOSKELETAL CARE?

Good musculoskeletal care includes proactive management of potential problems and a daily focus on range of motion exercises, positioning, and/or bracing. It should also include care by a team of MSK specialists.

WHO SPECIALIZES IN SMA MUSCULOSKELETAL CARE?

Team members can vary depending on the clinic set-up but may include occupational therapists, physical therapists, pediatric orthopedic surgeons, pediatric rehabilitation physicians, and orthotists. Occupational therapists (OT) focus on functional activities such as feeding, dressing, and other skills that require the use of the arms and hands. They may recommend and make splints and teach exercises to maintain range of motion and improve function. They may also recommend equipment to help with some of the above skills and activities.

Physical therapists (PT) focus on the legs and feet and functional activities such as helping a child move around in their environment. They may recommend orthoses, and teach exercises to maintain range of motion and improve function. They may recommend equipment such as special seats, strollers, and wheelchairs.

Pediatric orthopedic surgeons focus on surgical interventions for MSK problems, while pediatric rehabilitation physicians (also referred to as physiatrists) focus on nonsurgical approaches to MSK problems. Orthotists focus on making and adjusting orthoses.

Your MSK specialists should have experience in the care of children with SMA. This may not be possible for many children living in rural areas. If not possible, than an MSK specialist should seek direction from specialists that do work with SMA children.

HOW OFTEN SHOULD YOU SEE YOUR MSK TEAM?

Children should be seen by their SMA team at least twice yearly, and the team should include someone who specializes in MSK issues. Children may see a PT and/or OT in their community or school in addition to the PT and OT at the SMA clinic. The frequency of the therapy can vary but may be necessary as often as weekly when children are younger. If a child is having MSK problems, he or she may need to see MSK specialists more frequently.

Contractures in Children with SMA

DEFINITIONS:

AFO: ankle-foot-orthosis, which is an L-shaped splint that keeps the foot and ankle in a flat position.

Orthoses: custom-made braces used to align, support, prevent, and correct joint positions. They sometimes are referred to as splints, especially when used in the wrists or hands.

Range of Motion (ROM): the movement around a joint.

Contractures: fixed tightness around joints, which limits range of motion.

WHAT SHOULD YOU KNOW ABOUT CONTRACTURES?

Range of motion is maintained in the joints by moving them. A person can do their own range of motion by moving their limbs or having someone else move the limbs. Children with SMA have a difficult time moving their arms and legs due to weakness. This decrease in movement leads to the development of contractures. Contractures are fixed tightness around joints, caused by abnormal shortening of muscle tissue

Contractures can develop even when a child has some movement in their limbs. Contractures can also develop even when someone else is doing the range of motion on the joints. Contractures are most commonly found at the ankles, knees, hips, elbows, and wrists.

WHAT ARE THE GOALS OF ADDRESSING CONTRACTURES?

The overall goal is to prevent the development of contractures. If contractures are already present, the goal is to keep them from getting worse.

HOW ARE CONTRACTURES ASSESSED?

Contractures can be assessed by determining the range of motion of the child's limbs. Range of motion (ROM) is measured by moving the limbs to see how much flexibility they have. A tool called a goniometer (special ruler) can determine the ROM at all of the joints.

WHAT ARE THE DIFFERENT TREATMENT OPTIONS?

Range-of-motion activities can be performed on all of the joints in the arms and legs. These exercises can de done in therapy at a gym, in water therapy, or by stretching at home. Positioning the feet in a proper position in the wheelchair can also be helpful.

Splints or orthoses can be used to maintain, or sometimes even improve, range of motion. A cast can be applied and changed frequently to improve range of motion (serial casting). This is mostly done at the ankles. Surgery is occasionally, but not frequently, recommended to improve range of motion.

IS THERE SPECIAL EQUIPMENT THAT CAN HELP AVOID CONTRACTURES?

A stander can be used to maintain flexibility at the hips, knees, and ankles. Splints and orthoses can be used on both the arms and legs. Wheelchair footplates and leg rests should be positioned appropriately.

WHAT ARE THE SPECIAL CONSIDERATIONS FOR THE DIFFERENT TYPES OF SMA?

Non-sitters

For non-sitters, gentle range of motion of the arms and legs can be performed. A physical or occupational therapist can help teach these exercises. Splints for the hands can be used to maintain flexibility.

Sitters

Daily stretching activities can be taught by a physical or occupational therapist and should include elbows, wrists, fingers, hips, knees, and ankles. Standing in a stander can help provide a stretch on the hips, knees, and ankles. Lying on the stomach provides a stretch on the hips. Wearing an ankle-foot orthosis (AFO) as much as possible will maintain flexibility at the ankles.

Standers/Walkers

Maintaining the child's ability to walk as long as possible will help maintain range of motion at the hips, knees, and ankles. Wearing AFOs at night will help maintain flexibility at the ankles.

Bone Health in Children with SMA

DEFINITIONS:

Bone mineral density (BMD): a test that measures the density of minerals (such as calcium) in your bones using a special x-ray called a DEXA. This information is used to estimate the strength of bones.

Computerized tomography (CT scan): specialized x-ray study that looks at different views of a structure, which gives more detail than a regular x-ray.

Duel Energy X-ray Absorptiometry (DEXA): an x-ray that measures the amount of bone mineral content (BMC) in the femur, spine, radius, or whole body.

Osteopenia: mild to moderate decrease in bone mineral density.

Osteoporosis: severe decrease in bone mineral density that is associated with a higher chance of a fracture.

Fracture: a bone that is broken.

Z-score: the result of the DEXA is referred to as a z-score and is the measure of how close or far the result is from that of the average child of the same sex and age.

WHAT SHOULD YOU KNOW ABOUT BONE HEALTH?

Children's bones get stronger as they walk and perform physical activities such as jumping and running. Children continue to gain bone strength into early adulthood. Children who have physical disabilities may have weak bones.

Children with SMA may have decreased bone mineral density (bone strength) because of weak muscles and limited walking. A child who has decreased bone strength can develop osteopenia or osteoporosis. Children with decreased bone strength are at higher risk of having a broken bone.

Broken bones in children with SMA are often not related to trauma. They can be caused by minimal or no trauma and can even be due to getting a leg caught when doing a simple transfer. The leg is a common place to have a broken bone but it can occur in any bone.

WHAT ARE THE GOALS OF ADDRESSING BONE HEALTH?

The overall goals are to prevent fractures in children with SMA and to maintain strong bones.

HOW IS BONE HEALTH ASSESSED?

Checking vitamin D levels in the child's blood can assess bone health. A DEXA scan or CT scan can measure the amount of mineral content in the bones.

WHAT ARE THE DIFFERENT TREATMENT OPTIONS?

There are several different ways to try to prevent fractures from occurring. However, fractures can still occur even when all of the suggestions outlined below are followed.

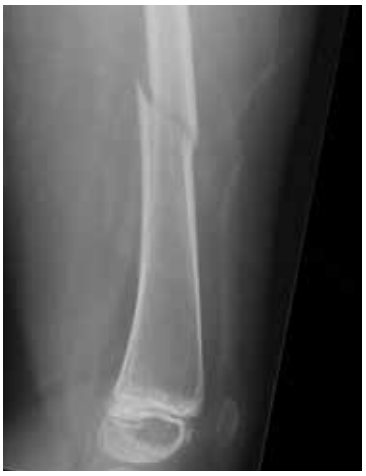
Recommendations include active weight-bearing exercise such as walking in those that can stand and/or walk. Passive standing in a stander may improve bone strength. The amount of standing that is needed is not known but may be greater than seven hours per week.

If vitamin D levels are low, supplements can be provided. The SMA team or pediatrician can determine what level is needed. Adequate calcium in the diet is also important, and should be supplemented if necessary. If the child has one or more fractures not related to trauma, medications called bisphosphonates may be considered. These can be prescribed by different specialists including the SMA doctor, an orthopedic doctor, or an endocrinologist.

Caution should be used when moving a child's arms and legs. Always provide support to the limb, keep the child's own alignment, and avoid the limbs being in a dangling position.

IS THERE SPECIAL EQUIPMENT THAT CAN HELP WITH DEVELOPING BONE STRENGTH?

Children may use a stander to help them to stand. They can use it for two hours a day, five days a week to maintain bone strength. There is also a wheelchair that helps position a child in the standing position.



Fracture

WHAT DO YOU DO IF A FRACTURE DOES OCCUR?

Most fractures can be treated with splints or casts. Some children don't need to be in a cast if they do not walk. Those children can be put in a soft splint. Casts and splints should be very lightweight, and they should be discontinued as soon as the fracture is healed. Sometimes when a child has a cast on his or her leg from a broken bone, a special wheelchair can be rented that allows his or her leg to be out straight.

A child who breaks his or her bone may need to have it fixed in surgery. An orthopedic surgeon will decide how to fix the broken bone. If a child needs surgery, it is important for all of the child's doctors to know that he or she has SMA. They can make sure other potential issues, such as breathing and nutrition, are taken care of. If a child who walks is put in a leg cast, it is important to get them back up and walking as soon as the cast is removed and the doctor says it's okay. This can help prevent any further weakness from developing. Some surgeons will recommend surgery rather than casting in order to get the child up walking more quickly.

WHAT ARE THE SPECIAL CONSIDERATIONS FOR THE DIFFERENT TYPES OF SMA?

Non-sitters

Special care should be used when performing ROM exercises. Supplemental calcium and vitamin D may be given to maintain high levels of vitamin D in the blood. If a child has one or more fractures, they may benefit from a bisphosphonate medication that can strengthen the bones. Standing in a stander may be very difficult because of severe weakness in the child's head and torso, as well as tight hips, knees, and ankles. An infant's respiratory status or general health may make standing difficult.

Sitters

Special care should be used when performing ROM exercises. Supplemental calcium and vitamin D may need to be given to maintain high levels of vitamin D in the blood. If a child has one or more fractures, they may benefit from a bisphosphonate medication that can strengthen the bones. Standing in a stander may be very difficult because of tight hips, knees, and ankles.

Standers/Walkers

Special care should be used when performing ROM exercises. Supplemental calcium and vitamin D may need to be given to maintain high levels of vitamin D in the blood. If a child has one or more fractures, they may benefit from a bisphosphonate medication that can strengthen the bones. Children may use a walker or a gait trainer to help them to walk. Maintaining the child's ability to walk as long as possible will help maintain bone strength. Active weight-bearing activities help improve bone strength. A child who is falling frequently may be at risk for having a broken bone.

The Hips in Children with SMA

DEFINITIONS:

Abduction: if hips are abducted, they are spread away from the body.

Contractures: decreased range of motion around a joint due to tightness in the muscles, tendons, or ligaments.

Dislocation: the top of the femur bone is out of the hip socket.

Dysplasia: shallow hip socket (acetabulum).

Subluxation: femoral head is not covered completely by the hip socket.

Osteotomy: surgery where the bone is cut and the alignment is changed.

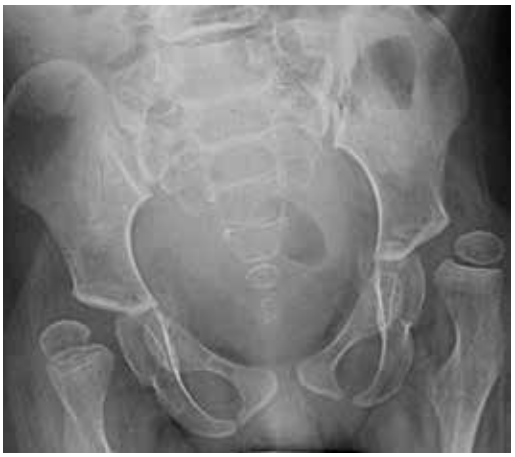
WHAT SHOULD YOU KNOW ABOUT THE HIP?

The hips are typically normal at birth. Hip development is adversely affected by weakness, imbalance of muscle (strong pull of some, weaker pull of others) and absence of weight bearing (standing).

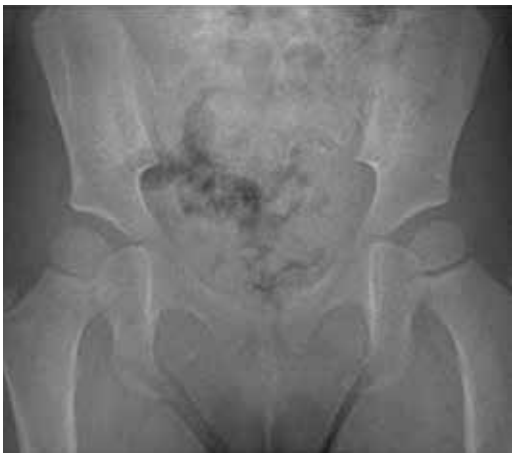
In children with SMA, several changes can occur. The femur (thigh bone) can become straighter than normal at the hip (coxa valga). The socket (acetabulum) can grow shallow and lose its cup shape. The femur can gradually "wander" from the socket, until it is no longer covered. A pelvic tilt (obliquity) can lead to an increasing hip deformity.

A hip deformity can be shown by a loss of motion. There may be decreased abduction, meaning the legs spread open less. The muscles in front of the hips may develop tightness. Legs may both point to the same side (windblown).

Feeling or hearing a "click" or "clunk" of the hip may represent that it is moving in and out of the socket. A dislocated hip can become very painful and make sitting difficult.



Hips Out of Socket



Hips in Socket

WHAT ARE THE GOALS FOCUSING ON THE HIPS?

Ideally the goal is to keep the hips in the socket. The focus is to have minimal or no hip pain in the long term.

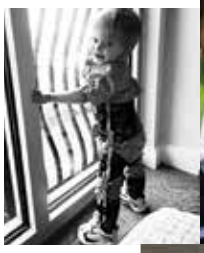
HOW ARE THE HIPS ASSESSED?

The hips are assessed by physical examination during every visit. An orthopedic surgeon or one of the SMA doctors may perform the exam. An x-ray of hips can be taken if there is concern for a hip problem.

WHAT ARE THE TREATMENT OPTIONS FOR MANAGEMENT OF THE HIP?

There are no known well-researched non-surgical ways to prevent the hip from subluxating or dislocating. One option to consider is correct positioning in the wheelchair or stroller with continued review and adaptation as the child grows. Physical therapy may help maintain range of motion at hips and promote weight bearing. Positioning the legs in an abducted position using a brace, pillow, or foam wedge may also be beneficial.

WHAT SURGERIES CAN BE RECOMMENDED IF THE HIP IS SUBLUXATING OR DISLOCATING?



There is no consensus on whether early attempts should be made to keep the hips in the socket. There is also no consensus on whether to recommend surgery to put the hips back in the socket if they are subluxated or dislocated. It is important for you to discuss the benefits and risks of any surgery with your SMA doctor and orthopedic surgeon.

In muscle and tendon surgery, a tendon can be lengthened or released with the goal of decreasing the contracture and rebalancing the force of the muscles.

Bone surgery (osteotomy) redirects the femur into the socket. It may include the use of hardware such as pins, plates, and screws. Bone surgery may require a bone graft.

Hip socket surgery (acetabuloplasty) is performed to rebuild the hip socket. There are different types of procedures depending on the age of the child and the status of the hip socket. Resection of the top of the femur may be necessary if the hip cannot be reconstructed.

WHAT ARE THE SPECIAL CONSIDERATIONS FOR THE DIFFERENT TYPES OF SMA?

Non-sitters

For non-sitters, it's important to maintain correct positioning in the wheelchair or stroller. Flexibility can be maintained at hip joints by performing range of motion exercises. Standing with support can be attempted, as long as it is tolerated. Surgery may be considered to redirect the femur into the hip. The surgeon should be made aware of the child's respiratory status and the pulmonologist should be involved pre-surgery.

Sitters

Correct positioning is also important for sitters. Flexibility can be maintained at hip joints by performing range of motion exercises. Standing with support can be attempted, as long as it is tolerated. Surgery may be considered to redirect the femur into the hip. The surgeon should be made aware of the child's respiratory status and the pulmonologist should be involved pre-surgery.

Standers/Walkers

Flexibility can be maintained at hip joints by performing range of motion exercises. Standing with support can be attempted, as long as it is tolerated. Surgery may be considered to redirect the femur into the hip, if the hip is painful. Surgery may be considered to rebuild the hip socket. The surgeon should be made aware of the child's respiratory status and the pulmonologist should be involved pre-surgery.

The Spine in Children with SMA

DEFINITIONS:

Scoliosis: spinal curve to the side.

Kyphosis: spinal curve forward (hunchback).

Lordosis: spinal curve of the low back inward (swayback).

Flexible: the spine is curved but can be straightened out with positioning or pressure.

Rigid: the spine is curved but cannot be straightened out, even with positioning or pressure.

Spine fusion: a surgery done to make the spine straighter, usually using metal rods.

VEPTR (Vertical Expandable Prosthetic Titanium Rib) System: hardware that is surgically implanted in younger children with severe scoliosis that allows for growth of the spine through periodic adjustments for growth.

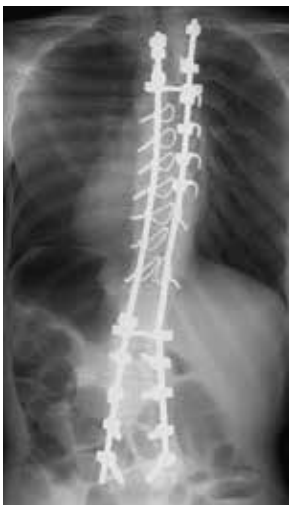
Growth Rods: hardware that is surgically implanted in younger children with severe scoliosis that allows for growth of the spine through periodic adjustments for growth.

TLSO (Thoracic-Lumbar-Sacral Orthosis): body jacket, also known as a brace that provides external support to the spine.

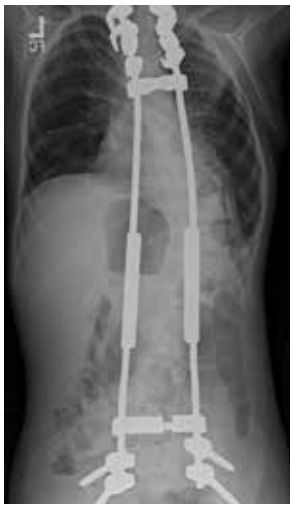
WHAT SHOULD YOU KNOW ABOUT THE SPINE?

Children with SMA are at high risk for a spinal curve because of the weak trunk muscles. Early spine deformities can affect lung development. Children who use wheelchairs are at higher risk for developing a spinal curve than children who walk.

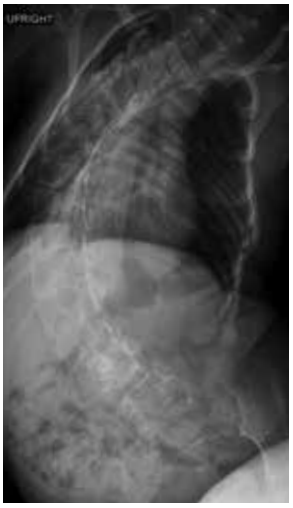
A curved spine can make it difficult to do different activities including sitting and using the arms (because a child tends to lean on one arm). There may also be problems with feeding/swallowing and breathing. A spine curve can cause pain with sitting or pressure sores. Orthopedic surgeons are the specialists who focus on the spine.



Spine Fusion



Growing Rods



Scoliosis

WHAT ARE THE GOALS FOCUSING ON THE SPINE?

The overall goal is to prevent a spinal curve or at least minimize progression of a spinal curve. A level pelvis should be maintained, along with an optimal sitting position.

HOW IS THE SPINE ASSESSED?

The spine should be examined at each clinic visit. An x-ray should be obtained if there is any curve noticed during the exam. Repeat x-rays may be obtained every 6-12 months to follow the progression of the curve once it has been identified.

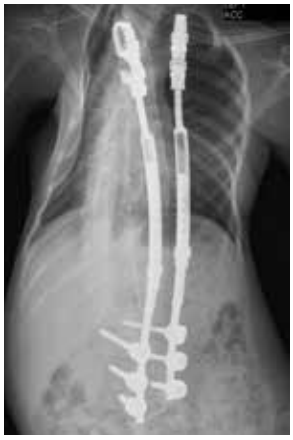
WHAT ARE THE TREATMENT OPTIONS FOR MANAGEMENT OF THE SPINE?

There are no known well-researched non-surgical ways to prevent the spine from developing scoliosis. To keep the spine straight, correct positioning while sitting should be practiced from an early age. Specialized components can be added to a wheelchair or specialized stroller, such as a custom seat back, lateral chest supports, and shoulder or chest straps. A TLSO can also be used to help keep the flexible spine straight. There is no evidence that it will stop the curve from progressing but it can be very helpful in keeping the child upright in their wheelchair.

Wheelchair and seating modifications can also be considered. This may improve balance and posture, leading to more comfortable sitting. It can also permit the arms to move more freely, thus improving function.

WHAT SURGERIES CAN BE RECOMMENDED IF THERE IS A SEVERE SCOLIOSIS?

The overall goal of surgery is to prevent the curve from getting worse, make the spine straighter, and make sitting more upright. Surgery can also give the lungs and internal organs room to grow and fully develop. The timing of a spine surgery depends on many factors including the age of the child, their respiratory status, and the rate at which the spine is progressing.



VEPTR

The two general types of scoliosis surgeries are 1) growing constructs and 2) definitive spine fusion. Growing constructs provide internal support, allow for ongoing growth of the spine, and allow the chest to continue to grow. They may require periodic surgical adjustments over time to allow for growth. Growing constructs include growing rods, in which the hardware attaches from the vertebrae to another vertebrae, and VEPTR, in which the hardware attaches from the vertebrae and pelvis to the ribs.

Spine fusion surgery permanently fuses the vertebra. Hardware is attached to each vertebra and often goes into the pelvis. No motion or growth can occur over the levels that the spine is fused. Bone graft is commonly used to aid in fusing the separate vertebra together over 6-12 months. The fusion process eliminates further growth of the spine, which subsequently limits the risk for further progression of the scoliosis.

WHAT ARE THE SPECIAL CONSIDERATIONS FOR THE DIFFERENT TYPES OF SMA?

Non- sitters

Maximum support should be provided using specialized seating. A TLSO can be tried but it may be difficult to fit and can interfere with breathing. Depending on the medical status and age, a growing construct can be considered in a child with a very severe curve in order to allow ongoing growth of the spine.

Sitters

Maximum support should be provided using specialized seating. A TLSO can be tried but it may be difficult to fit and can interfere with breathing. A growing construct can be considered in a child with a very severe curve in order to allow ongoing growth of the spine. A definitive spine fusion can be considered in an older child or teenager with a severe curve. The surgeon should be made aware of the child's respiratory status and the pulmonologist should be involved pre-surgery.

Standers/Walkers

A TLSO should be tried but could interfere with walking. A TLSO will likely not interfere with breathing in this case. A definitive spine fusion can be considered in an older child or teenager with a severe curve. Fusing into the pelvis in someone who walks may make walking more difficult. The surgeon should be made aware of the child's respiratory status and the pulmonologist should be involved pre-surgery.

Conclusion

In summary, children with all types of SMA are at risk for many MSK complications that stem from the underlying weakness and low tone. Having a team of specialists supporting your child from a very young age will be important. Non-surgical treatments including stretching, positioning, orthoses, and equipment may be helpful. Some children, even when provided with intensive interventions from an early age, go on to develop complications that may at times need surgery. The ultimate goal is to minimize MSK complications as much as possible and maximize your child's functional abilities and overall health.

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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