



# Joshua Frase FOUNDATION™

## FAMILY GUIDE TO MYOTUBULAR MYOPATHY

**A starting point for those new to the MTM Community.**

The diagnosis of Myotubular Myopathy (MTM, or XLMTM) can be scary and overwhelming. We hope you take comfort in the fact that others have experienced what you are going through. Welcome to one of the strongest community support groups of any rare disease! The parents and families of children with MTM are united in helping each other and are working together to find the first treatment for this disease. We invite you and your family to connect with us in whatever way you can and we look forward to getting to know you. The next few pages will contain vital information to take you from birth to caring for your child at home, and beyond...



### **DISCLAIMER**

*The information and advice published or made available in this booklet is not intended to replace the services of a physician, nor does it constitute a physician-patient relationship. This advice should be taken in conjunction with medical advice from your medical clinician whom you should consult in all matters relating to your child's health, in particular with respect to symptoms that may require diagnosis or medical attention. Any action on your part in response to the information provided in this booklet is at your own discretion.*

## HOW TO USE THIS MTM WELCOME PACKET

### From the NICU to the first months at home

The purpose of this guide is to give the most basic information to new families and to help connect the newly-diagnosed with available resources. This document was created in 2013 by the Joshua Frase Foundation Educational Advisory Board, which consists of informed parents whose families have been living with this disease. This document was also reviewed by the Joshua Frase Foundation Scientific Advisory Board. We have created this welcome packet to assist new families in handling the large amount of medical information that they are encountering at the start of this journey. We understand that there is a wide range of severity for people with MTM. Not all items may apply to your case. This document may also be useful to families with severe forms of Centronuclear Myopathy (CNM) or other similar neuromuscular diseases.

Unfortunately, you will encounter physicians that have never heard of MTM, or if they have heard of it they may only have outdated and somewhat bleak information. We want to be clear, this is not your doctors' fault, MTM is a rare orphan muscle disorder that few have heard of. We urge you to share this guide with health care providers, family members, school staff, and/or other care providers. Your doctor may be able to help explain some of the points made in this guide if you have questions. Additionally, there is a recent (2012) article published in the Journal Of Child Neurology which was the result of collaboration among several medical professionals and provides a general overview of the Standard of Care for Congenital Myopathies, including Myotubular Myopathy. This article, the [Consensus Statement on Standard of Care for Congenital Myopathies](#), should be shared with your medical team. The Joshua Frase Foundation website has links to this and other resources which may be helpful for your medical team under the "[For Clinicians](#)" section. The Standard of Care is a more technical document concerning medical management for a variety of Congenital Myopathies. Our document is intended to give a brief overview specifically focusing on MTM management in an easy to understand format.

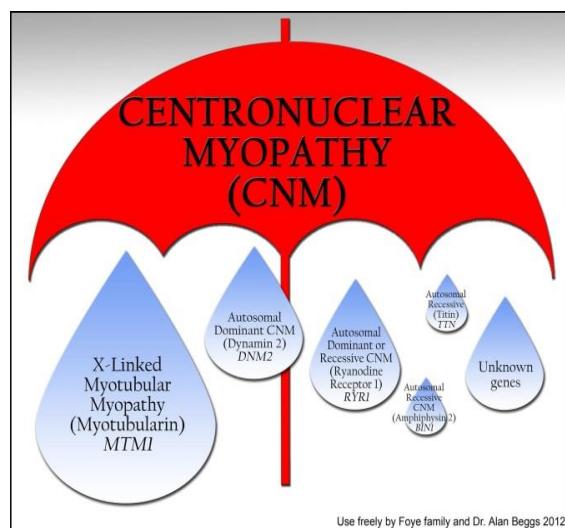
### How you arrived here

If you are reading this you probably have a child that was born "floppy." Your doctor may have told you that your child may have a myopathy and you scoured the Internet for myopathy information. Your child may have had a muscle biopsy and you have been told that your child has Centronuclear Myopathy or Myotubular Myopathy. Finally, your child may have undergone a genetic test to confirm a diagnosis of Myotubular Myopathy. If you fall into any of these categories this guide may help you and your physician.

### Centronuclear Myopathy v. Myotubular Myopathy

The term Centronuclear Myopathy is a broad term that includes several different specific myopathies that share a common "look" when viewing the muscle cells under a microscope. This umbrella shows the different conditions that fall within Centronuclear Myopathy.

There are more kids with MTM than any of the other myopathies under the CNM umbrella, and at one time the terms MTM and CNM were used to mean the same thing. Today, the term Myotubular Myopathy is still sometimes misused as a broad term for CNM when in fact the only way a person can be officially diagnosed with Myotubular Myopathy is with a genetic test. A muscle biopsy can lead to a diagnosis of Centronuclear Myopathy, but genetic



testing is needed to determine the particular sub-type. If you have not had genetic testing, it is recommended that you get genetic confirmation. This is important because your child may have one of the other myopathies as shown in the umbrella diagram.

### **Confirmation of MTM is done by a genetic test**

In order to get a genetic confirmation of MTM, a small sample of blood is sent to a specialized genetic testing laboratory. The University of Chicago has set the standard for CNM/ MTM testing. [Download form here](#). For other comparable testing laboratories worldwide, [please click here](#). If you have not obtained genetic confirmation, your physician should be able to have blood drawn and sent to the laboratory at the University of Chicago for testing. Although most insurance and state-funded health care plans will pay for this test, there are programs available that may pay for this test if you are unable to get testing done through your medical coverage provider. *Once you have genetic confirmation, hold on to your original documents like you would a passport.* You WILL need to show the genetic report to researchers if you choose to take part in clinical trials for new drugs and therapies.

### **WHAT IS MTM?**

Myotubular Myopathy (XLMTM or MTM) is a rare genetic disorder. This particular disorder affects the strength of the skeletal muscles. The severity of the disease differs from case to case, but is usually life-threatening. Most often, MTM will be present at birth, and a child born with this condition is likely “floppy”, or in scientific terms show signs of “hypotonia” – which means “low muscle tone”. Your baby may be extremely weak. Most of the time, these babies will also struggle with the ability to suck, swallow, and breathe on their own because all of these basic tasks use muscles. Generally brain development and intelligence is not affected by MTM, nor is the heart function directly affected as the heart is a different kind of muscle. Despite the challenges of MTM, there are many individuals around the world living and thriving with the disease, thanks to improvements in medical technology and care. There are some very exciting research programs underway that hold great hope for improving muscle strength in MTM in the future.

### **GENETICS AND DIAGNOSIS**

MTM is a genetic disease, which means that the person with MTM is born with it in their genes, much in the same way they are born with a particular hair color or body type. Often, a change (or mutation) in this gene is passed down from the mother ([x-linked overview in genetics](#)) , though the mother is usually not affected by the disease in the same way a boy is, due to the differences in chromosomes between males and females. Chromosomes are the packages of genes. The main difference between males and females are the sex chromosomes, X and Y. Males have one X and one Y chromosome, whereas females have 2 X chromosomes. The gene for MTM is located on the X chromosome. Since males only have one X chromosome, they will show symptoms of MTM, while females have 2 X chromosomes which explains why they may show milder or no symptoms. Usually the mother does not even know she carries a change in the gene until an affected child is born. Sometimes a boy is born with MTM due to a spontaneous change in the gene, meaning the mother is not a carrier. Understanding the genetics of MTM can be important for several reasons, and it is recommended that you contact a good genetic counselor (such as at the [Beggs Laboratory](#), also see references below) to discuss getting genetic confirmation.

### **BECOMING YOUR CHILD'S ADVOCATE AND BUILDING YOUR CARE TEAM**

The person who will lead the management of your child's care is you, the parent. Because MTM is so rare, many doctors and hospitals have little if any experience diagnosing and caring for someone with MTM, and the initial doctors you encounter (for example, in the NICU) may not know how to deal with your child. It will end up being your responsibility to make sure you have a great team of doctors and therapists. Do not be afraid to get second

opinions and look for new doctors if you are not satisfied with the care you are receiving. Advocate for what we know you will need, as detailed in the next sections. You should have on your team a Pulmonologist, a Neuromuscular Specialist, Gastrointestinal Physician, Pediatric Orthopedist/Orthopedic Surgeon, Physical Therapist, Occupational Therapist, Speech Therapist, Rehabilitation Specialist (Physiatrist) and hopefully a very good primary physician that can help coordinate the care of all of these specialists.

A good resource for physician recommendations is the other parents. They have invaluable experience and advice concerning the care of MTM children. The Facebook group "[MTM/CNM Family Support & Discussion](#)" is a meeting place for MTM parents. Members will answer questions and help you in any way they can. It's also a place where you're surrounded by people who 'get it' - which is a priceless resource.

## BREATHING AND RESPIRATORY CARE

The main physical effect of MTM is weakened muscles. The weakened breathing muscles in particular (diaphragm and chest muscles) lead to the primary day-to-day concern in MTM: breathing difficulties. Most severe illnesses and deaths in MTM are the result of some sort of breathing complication.

### **MANAGING YOUR CHILD'S BREATHING IS THE #1 PRIORITY FOR MOST MTM CHILDREN.**

Managing respiratory rates, oxygen levels and secretions is a daily life and death battle. The key to your child's long term health is to avoid pulmonary complications such as mucus plugs, pneumonia, and collapsed lungs. Do not despair, with proper airway maintenance many problems can be avoided, and you will learn to deal with these types of issues as a normal part of taking care of your child. A large percentage of MTM kids will benefit from a breathing machine, such as a ventilator, that will help blow air into their lungs. Ventilators take over much of the work that the breathing muscles normally do, which relieves some of the stress on these weakened muscles. This is called mechanically-assisted ventilation, and is described in more detail below.



### **Finding the right pulmonologist and why this doctor is important**

It is essential to find a doctor who has experience with managing respiratory care and mechanically-assisted ventilation in an individual with MTM or other severe neuromuscular weakness. Often this doctor is a pulmonologist (lung doctor), though not every pulmonologist has expertise in working with people who have neuromuscular weakness. Many NICU pulmonologists are used to dealing with premature infants, in which the goal is to gradually wean the child from support as the lungs mature. However, since the breathing issues in MTM are primarily related to muscle weakness rather than premature birth (though prematurity is often seen in MTM as well, which can complicate the situation particularly prior to diagnosis), the plan will look very different for a child with MTM. Other pulmonologists specialize in diseases of the lung such as Cystic Fibrosis. In MTM, the lungs are generally "healthy" - they just need help with the mechanics of breathing and staying clear of secretions and mucus. Your Pediatric Pulmonologist will often become the most important physician for the long term care of your child.

Sometimes, this important role will be played by a doctor other than a pulmonologist. For example, some Ear, Nose, and Throat (ENT) or Oto-Rhino-Laryngology (ORL) doctors specialize in tracheostomy and mechanically-assisted breathing in patients with neuromuscular issues. Some hospitals have developed multi-disciplinary tracheostomy teams for both inpatient and outpatient care. These multi-disciplinary teams are often led by a Hospitalist (a doctor who specializes in intensive care), who can be an excellent primary resource for a patient with

MTM. It is important that you take an active role in seeking out the best available resources for respiratory management at your hospital.

## BREATHING MACHINES

### The types of medical equipment often used to help in breathing

There are several types of machines available that help to blow air into the lungs of a person with MTM, which helps them to take the breaths they need even if their muscles are too weak to do this on their own. In some less severe cases, this machine might be needed only occasionally, or perhaps only while sleeping, when the natural drive to breathe is low. In these less severe instances, the air is usually delivered to the patient through a mask, which can cover the nose or both the mouth and nose. An advantage of mask-delivered assistance is it (also called “non-invasive ventilation”) can be removed when it is not needed. However, most cases of MTM tend to be severe. Many children will require breathing assistance for most or all of the time, and they will benefit from a more permanent option. This is usually in the form of a tracheostomy or “trach” tube, which is a small, flexible tube that is placed through the front of the neck into the airway by a surgeon. The trach provides a direct link between the ventilator (breathing machine) and the lungs and actually makes daily management of breathing issues easier to deal with in many ways.

### Tracheostomy and Ventilator

It is generally agreed that having a tracheostomy allows for easier access to the airway with suction catheters and allows for the better removal of secretions that can cause mucus plugs. It is also generally agreed that a ventilator allows the child to develop in other ways, since the child is not using so much effort and energy to breathe. If your MTM child requires a significant amount of oxygen, has been placed on BiPap or has been placed on CPAP then it is strongly recommended that you consider a tracheostomy and ventilator. (See note below concerning CPAP). When deciding whether to use mask-delivered or invasive ventilation, discuss the options with your child’s care team, including the pulmonologist, primary care physician, and therapists. Consider the amount of time your child needs assistance, his ability to manage the secretions from his mouth, and whether the mask interface obstructs your child’s ability to interact with the environment. It is difficult to find an MTM parent with a child on a ventilator who regrets the decision to have the tracheostomy and the ventilator support, and most have reported a significant increase in their child’s overall wellbeing and quality of life.



Today’s ventilators are quite advanced and relatively portable, allowing our kids to enjoy school, travel, and a full range of experiences with some extra planning and preparation.

Some of the negative effects of the tracheostomy/ventilator are the dependence on the machine and a reduced capability for speech. Although some children learn to speak, others may not.

Most of these risks can be alleviated through proper trach care and early intervention with a speech therapist, who will be vital in training your child to speak with while having a trach. Some individuals with MTM also communicate with sign language or assistive communication devices.

*Continuous positive airway pressure (CPAP): CPAP is a treatment that uses continuous air pressure to keep the back of the throat open. It is designed for people with sleep apnea only. It will not treat hypoventilation due to weak respiratory muscles. It is not appropriate for people with neuromuscular conditions because it does not provide enough assistance for breathing in and it actually makes breathing out even harder for them. People that have strong breathing muscles and need treatment for apnea could benefit from CPAP use.*

## **OTHER MEDICAL EQUIPMENT COMMON TO MTM HOMES**

If you are leaving the hospital without the following equipment, you should ask your doctors why your child does not need them. While MTM has varying degrees of severity, these pieces of equipment are very important to almost all in the MTM community.

### **Suction Machine**



This is a vital piece of equipment that you should likely have with you at all times. The suction machine is essentially a vacuum for mucus. This machine helps clear out mucus and secretions from the airway. Muscle weakness MTM affects swallowing and therefore the suction machine is also used to remove saliva and mucus that accumulate in the mouth and nose. The buildup of saliva and mucus can be a breeding ground for infections and can cause mucus plugs. A mucus plug is a thicker secretion that the child is unable to cough up due to the muscle weakness.

Mucus plugs can be fatal as they can block your airway and prevent air from passing in and out of the lungs. The process of keeping the air passages clear from mucus is called “pulmonary hygiene.” In addition to the suction machine, the other equipment can help you monitor and maintain your child’s daily pulmonary hygiene.

### **Pulse Oximeter**



Commonly called a “Pulse-Ox” or “Sat Monitor”, this machine is a must-have piece of equipment for all. This device monitors the oxygen levels in the blood (oxygen saturation) as well as the pulse rate. This monitor gives advanced warning when our children are in distress and their oxygen levels drop or heart rate changes. This is your primary warning if a mucus plug develops. You will quickly learn your child’s usual heart rate. Knowing this information will give you advance notice of impending illness, as the heart rate will likely increase before you see outward signs on your child’s face.

### **Nebulizer**

A nebulizer is a machine that allows certain liquid medications to be vaporized and inhaled into the lungs. Many of our children take medicines such as Albuterol, Pulmicort, Pulmozyme, DuoNeb and FloVent as part of a daily regimen of airway maintenance. These medications may act to reduce inflammation in the airways or to thin the lung secretions. The drugs are often given in the morning and again at night. Additional treatments are usually recommended when the child develops a common cold or other respiratory issue. It is believed that the survival rate of MTM patients is improved with aggressive airway maintenance.



### **Chest Physical Therapy**

Chest physical therapy (CPT) is used to loosen secretions. A percussive vest is often used as the primary therapy. There are many different terms and different brand names for a percussive vest, but in each instance, a machine uses air to rapidly inflate and deflate a vest. As a result, the vest causes shaking of the child’s chest. The same



therapy can be performed manually with percussion cups. The parent taps the percussive cups on the child’s chest repeatedly. CPT can be completed using either the percussive vest or cups. The vest, however, treats several different regions of the lungs at once, and further, it enables the parent to have free hands to manage suctioning and other tasks. Many parents believe that CPT has been vital to maintaining health and avoiding potentially fatal mucus plugs and pneumonias.

## **Cough Assist**

The cough assist also helps move the mucus up and out. It is an essential part of airway maintenance. The cough assist is a machine that mimics a cough. The machine connects to your child's tracheostomy tube if he is invasively ventilated, or it can be used with a mask in children who do not have a trach. First, it pushes air into the lungs to inflate them, and then sucks the air out. This sucking action mimics the forceful push of a cough, in attempt to move secretions out of the small airways of the lungs. Due to the muscle weakness, MTM children do not have strong coughs. In fact, many cannot cough at all. The cough assist machine is usually used in conjunction with the other treatments described above. The nebulizer treatment of Albuterol and Pulmicort is first given, followed by the percussive vest and then the cough assist. The cough assist is the final piece that helps clear the airway and lungs of the secretions that were loosened by the first two treatments. If your child does not have these three treatments as part of a daily regimen, it is suggested you ask your pulmonologist why you are not on a similar program.



## **Important Note Regarding Respiratory Equipment**

All of the essential respiratory equipment listed above requires electrical power to function. It is important to request a backup device for each of these pieces of equipment when possible (particularly the suction machine and ventilator), a battery backup option if available, and/or adapters or cords that can be used during travel in vehicles. These backup power sources are helpful when you begin to venture out into the community but also are vital in cases of power outages and equipment failures. Many families will also invest in backup generators for their homes and you may speak with hospital support staff to find out if your state offers assistance for these circumstances. Also, it is recommended that you contact your power company and let them know that you have a child who relies on "life sustaining equipment", which will allow them to prioritize restoring power to your house in the case of a widespread outage. Another step can be to notify your local paramedic squad to let them know you have a child with a ventilator. This helps them get to know who they might be called on in the future.

## **Oxygen**

Most of the time, proper airway maintenance and ventilator support will allow the person with MTM to breathe "room air" just like everyone else. But sometimes they will need a little extra oxygen (room air is only 21% oxygen), which can be provided via a cylinder of compressed gas, a smaller liquid oxygen system, or an electronic oxygen concentrator. The oxygen can be given continuously through the ventilator in varying amounts or can be administered through a nasal cannula or face mask, as appropriate. Oxygen requires a prescription from your doctor, and like other medications it should be used as instructed. Your doctor may instruct to you to use oxygen regularly or "PRN" (as needed), to keep your child's oxygen saturation in an ideal range. It is important to understand that an increased need for oxygen usually indicates respiratory problems that must be addressed - for example suctioning a mucus plug, or treating for pneumonia. Do not give oxygen without also attempting to address the underlying problem and seek advice from your doctor if your child needs more oxygen than usual.



## **Resuscitation "Ambu" Bag**

An ambu bag is a hand-held device that is used to manually provide breaths of air into the lungs of patients who are not breathing adequately on their own. For most MTM patients, this is a vital piece of equipment that should be with you at all times. By squeezing this self-inflating bag, it gives full breaths via the nose and mouth or through the trach. Often, you



will give breaths from the ambu bag while routinely suctioning or when your child needs a fuller or deeper breath. It is also important to carry with you if your child uses a ventilator in case the vent malfunctions, the ambu bag will be used to provide the needed breaths while backup help arrives.

## FEEDING AND GASTROINTESTINAL CARE

### Feeding

As most of these children do not have the muscle tone needed for oral intake, they receive the nutrition they need from liquid given through a tube. It takes work to chew and swallow food, which may fatigue your child before they can eat enough food to be nourished. Further, if your child has significant problems with breathing, he may have difficulty safely coordinating with work of breathing and swallowing. Your doctor may prescribe a particular formula or you may decide to make a homemade formula, using a special blender called a Vita Mix. A Vita Mix is a powerful blender which liquefies food so it can pass through a small tube. Our community has had great success with the Vita Mix but know that there are other comparable blenders available on the market. Making the decision to make your child's food yourself requires planning and a consultation with a dietitian to insure adequate nutrient and caloric intake.

### NG/G-Tube

A NG tube is an abbreviation for a nasogastric tube, which is a flexible feeding tube that passes through the nose and down into the stomach. This is usually for short-term use.



A G-Tube is a small surgically placed device, commonly called a "button", that enters directly into the stomach, and it is a more permanent option than the NG tube. It is used to allow a liquid nutrition to be given without the need for the person to swallow.

Swallowing requires many muscles working in a coordinated fashion, including a "hatch" that closes at the right time over the airway, keeping food out of the lungs. Some children learn to swallow their own secretions and eventually some food or liquids, but it can take time. A test should be done at a hospital, with an X-ray, to see how your child's swallow is functioning, before any food or liquids are offered. The test is commonly called a swallow study.

*Almost all MTM children need a G-Tube.* The G-Tube and accompanying feeding machine are easy to use and allow children to grow while limiting the threat of food entering the lungs.

### Nissen Fundoplication

In the most general terms, this surgical procedure tightens the top of the stomach to help prevent reflux and vomiting. The muscles that limit reflux are also weak in MTM children. Those with significant vomiting and/or reflux may require a Nissen. In many cases, a Nissen is performed at the same time as the G-Tube placement.

Sometimes, with or without a Nissen, medication may be required to help manage reflux. Your gastroenterologist will help determine what medications and surgeries will be most beneficial.

## ORTHOPEDIC CARE

Most kids with MTM are unable to walk (non-ambulatory) and require the use of a wheelchair. These children are also non-weight bearing which can cause their bones to be thinner and weaker than other children their age. The earlier you can engage your child in exercise and physical therapy/occupational therapy, the more you can increase

the likelihood of maximizing their physical potential and bone strength. Working on range of motion exercises can prevent muscle and tendon tightening from reduced movement.

Your hospital social worker will have information about Early Intervention programs in your area and connect you to physical and occupational therapists, who can aid in your child's physical progress.

At birth, some MTM kids have clubbed feet, where the feet turn inwards at the ankle due to low muscle strength. An orthopedic specialist will likely guide you through the varying treatment options which include casting, bracing and tendon release surgery. Many times children will wear AFO's (ankle foot orthotics) to help keep feet in alignment. Wearing AFO's is a painless way to keep growing feet in alignment.



It is common for children with MTM to be very tall for their age. In many cases, children exceed the height of 90% of other children their age.



As your child grows, you will also need your orthopedic specialist to watch for spinal problems, such as scoliosis (sideways curving of the spine). Scoliosis is fairly common among MTM children since their muscle growth cannot keep up with their bone growth, and it can be difficult to maintain proper posture. Some children will wear a TLSO (thoracic lumbar support orthotic, also known as a "body jacket") which is like a hard vest to support the trunk area. Bone density tests can be done to track how bone density looks over growth spurts. Early intervention and having a good orthopedic specialist on your team can make this process run smoothly.

## MOBILITY



As mentioned previously, most kids are non-ambulatory and require the use of a wheelchair. Children usually take very well to a power wheelchair (an electric powered wheelchair, controlled by a joy-stick or other device). It Offers self-directed mobility to children that cannot otherwise do so in their early childhood development.

In many cases, children at earlier ages can manage toys that assist or benefit mobility while also aiding in muscle development.



Getting insurance, state aid or other insurance providers to pay for a wheelchair can be a long process and can take many months. Begin applying for your child's first wheelchair at 6-9 months. Many children will start with a stroller type wheelchair.

## OTHER ISSUES AND CONSIDERATIONS

MTM is a very rare disease, and despite great advances in research over the last several years, much is still unknown about MTM. While muscle weakness is clearly the main issue that affects all with MTM, there are some other issues that have been experienced by some in our community. These conditions are not universal, but might be somehow related to MTM either genetically or as "side effects" of the overall condition.

### Liver Issues

Sadly, we have lost a few in our community due to severe/sudden liver bleeding. The technical term is "peliosis of the liver" (or peliosis hepatis). It is not clear if or how this may be linked to the MTM gene which mainly affects

muscles, but there is still so much unknown about this disease. Peliosis hepatis manifests as blood-filled cysts in the liver tissue which can rupture and cause severe internal bleeding. There are a few case reports (some published, some just shared in our community) going back to at least the 1990's, though it hasn't really been studied much in terms of what to do for detection, prevention, and treatment. It is a somewhat rare complication in an already rare disease. Imaging studies such as MRI, CT Scan, and Ultrasound can be done to look for signs of peliosis, and periodic screening should be considered by you and your doctor.

Other liver issues have occurred with some children with MTM, such as elevated liver enzymes, high bile acid or bile salt levels and calcium based gallstones. Your child's specialist may suggest a liver biopsy. This procedure is not without significant risks and should be considered only in light of what therapy may be added or changed as a result of what is seen from the biopsy. MTM children have died as a result of uncontrolled bleeding during liver biopsies. It is important to note that although lab tests can indicate that bleeding times are normal, some of the children have prolonged bleeding despite normal lab test results. Sometimes, abnormal liver function can be caused by medications, and often liver problems resolve on their own.

### **Seizures**

Some children with MTM have also experienced seizures. Seizures are defined as an excessive surge of electrical activity in the brain. They can be focal (in one area of the brain), multifocal (in many areas of the brain), or generalized (throughout the brain). Some seizure types include absence seizures (previously known as petit mal), partial seizures, and tonic clonic seizures (previously known as grand mal). The cause of the seizures in these children is not well understood, though sometimes they can result from prolonged lack of oxygen or serious infections. Uncontrolled seizures can lead to significant brain damage and even death.

Seizures can sometimes be hard to identify or easily missed. For instance, during an absence seizure, a child may stare blankly or kind of "zone out" and be unresponsive. This type is easy to miss because children with MTM are often nonverbal and weak, and can have extended blinking times. Absence seizures typically last seconds to several minutes. Grand mal seizures are more obvious with the stiffening and jerking of the body. This type can last several minutes to much longer.

Certain conditions like high fever, low blood sugar, illness, may or may not precede seizures activity. Some children may experience an "aura" prior to the seizure itself that may consist of a feeling that a seizure is coming, numbness, a strange smell, or a strange taste. After a seizure (postictal period), children may be confused, sleepy, have a headache, be unresponsive, or be upset for a period. You may see high or low heart rates, a decrease in your child's oxygen levels, sweaty skin, and/or eyes rolling back. If you suspect your child is having a seizure, you should call out your child's name and try to determine whether he/she is responsive. An EEG (electroencephalogram) test can help diagnose seizure. An EEG will only show current seizure activity or seizure susceptibility, it will not show past seizure activity. In some cases, a prolonged EEG recording may be helpful. There is a lot of variability with seizures. If you think your child may be having seizures, speak with your healthcare provider.

### **Undescended Testicles**

Undescended testicles are common in MTM, and while not directly harmful it may increase the risk of testicular cancer later in life. Many parents have chosen to have the testicles manually brought down through surgical intervention.

## Dry Eyes

Dry eyes are extremely common as many MTM children do not blink completely or frequently enough. A nighttime ointment and daytime drops are used to keep the eyes moist and comfortable.

## Ear Infections

Fluid build-up behind the eardrums can occur due to lack of swallowing and may be linked to chronic ventilator use. This fluid can become infected, causing an earache. Some children have small ear tubes placed in their ear drums to allow excess fluid to escape, which helps to prevent ear infections.

## Bone Issues

Over time, bones that are not subject to weight-bearing exercise do not develop the same strength and thickness as active children. This can mean that bones are more susceptible to fractures. Your child's physician may refer you to a specialist to follow this. In general, calcium and vitamin D supplementation can be helpful in maintaining proper bone density and overall bone health.

## Other Complications

There are other complications that have been documented and/or self-reported and shared within our community. These complications have included: kidney stones, gallstones, spherocytosis (a change in the shape of the red blood cells), pyloric stenosis (a narrowing of the sphincter between the stomach and the small intestine), vitamin K-responsive bleeding tendency, delayed sexual maturity with low testosterone, dental issues due to a smaller mouth or high arched palate, hydrocephalus (an accumulation of fluid inside the brain), vision issues, hearing loss, low platelets, enlarged spleen, bleeding issues, slower processing speed for information, and low energy levels. When children with neuromuscular disorders receive anesthesia, they are at a slighter higher risk for complications related to anesthesia use. Generally anesthetics can be well tolerated, but it is very important that the medical professionals involved are aware of these higher risks.

*Please note: these are all RARE complications in a very rare disorder. Your child **may never** be affected by any of the 'other complications'. However, we want you to be fully equipped should the need arise.*

## **CARING FOR YOUR CHILD AT HOME**

Every parent needs to remember first and foremost, that MTM/CNM children are just that, children! They have all the hopes and dreams, drive, determination, love and caring of any other child born into this world. These MTM children are intelligent, loving, and downright funny!



They love to play games, do origami, build models, watch sports, drive go-karts, fly remote control planes, go for walks in the woods, play sports, hang out with a pretty girl, and even help out around the house and yard! Given as many of the same opportunities as any other child, MTM children thrive and love life!

In light of that, below you will find tips on how (and when) to put systems in place to help you give your child the best care possible.

### **Durable Medical Equipment (DME)**

The medical equipment common to MTM family homes is typically supplied and maintained by a Durable Medical Equipment (DME) Company. The best time to create your needed supply list is while you are still in the hospital with the team of medical professionals that know your child's needs. It is also the easiest time to get things approved by your insurance company as they will be anxious to discharge your child from the hospital to home care where care expenses will be less. Request all of the items that your team thinks you will need and ask for backup items as well. Sometimes the standard authorization provided by your insurance company will be less than what your child needs, as the standards are not always written with conditions like MTM in mind. Your doctor may need to write a Letter of Medical Necessity and/or a Letter of Overutilization to accompany the prescription and request that coverage is warranted for your child.

### **Nursing Care**

Some insurance companies and many state assistance programs provide for in-home nursing. The availability of nursing varies from state to state. Some states will provide in-home nursing only for those that are on a ventilator. The hospital social worker should be able to discuss nursing care with you and assist in selecting a Nursing Agency. The [Joshua Frase Foundation](#) can also help to connect you with other families in your area so you can learn more about your nursing options.

### **Early Childhood Intervention (EI) Therapies**

Early Intervention (EI) is a program that provides therapies to children with developmental delays. Early intervention services are federally mandated through the Individuals with Disabilities Education Act. Typically MTM children will be eligible for EI therapies until the age of three at which point the schools are required to take over therapies. Their EI therapies may include, but are not limited to; Physical Therapy (PT), Occupational Therapy (OT), and Speech Therapy. Other assistance offered through EI may also include assistive technology and nutritional services. Depending on the state many of these therapies will be provided in your home. These services may have a family fee depending on the state and your family's income.

### **Caring for the Caregiver**

The experience of having a special needs child is a daunting task and can sometimes feel overwhelming to you and your significant other. Know that it is normal and that reaching out for support from others such as family, friends, others in the community, and professional counselors is often helpful. No one is ever really alone. A family therapist can sometimes be helpful for families going through the changes associated with your special needs child. Remember that when you are physically and emotionally healthy, you can be a better caregiver for your child.

## **INSURANCE AND MEDICAID**

Medical expenses for individuals with MTM are extraordinary. Often, primary insurance plans will not cover all of the medical services that are needed and most do not include coverage for private duty home nursing that most children with MTM require. It is very important to look into what state programs that your child may qualify for including SSI (Supplemental Security Income) and/or Medicaid waiver programs. Every state has a Medicaid waiver program for children who depend on medical technology to live at home. In many states, our children with MTM will qualify for this Medicaid program that does not factor in a family's income, but rather is granted based on your child's medical needs alone. Although the intent of this waiver program was implemented specifically for children like ours, some states have exclusion clauses, waitlists, or limited resources. It is important to speak with hospital staff immediately to apply for whatever Medicaid waiver program your child may be eligible for and ensure the best coverage possible.

## **RESEARCH & FUTURE TREATMENTS**

Participation from our community is paramount to ensure the potential success of these therapies being studied. The research has advanced thus far because of families like you were willing to share their family and medical information with these scientists. To view all research accomplishments and research initiatives for our community, and to get involved please go to the [Get Involved](#) page on JFF's website.

There are two amazing biotechnology companies that are closing in on the first treatments for MTM. Audentes Therapeutics and Valerion Therapeutics are working on two different potential treatments which have shown great promise in the laboratory. Each company is moving closer to clinical trials (human tests) and they are very interested in connecting with the MTM community and making a difference for our kids. Check out their websites at: <http://www.audentestx.com/> and <http://valerion.com/>.

## **HONORING & REMEMBERING**

With the use of medical technology and advanced respiratory support, the number of individuals living and thriving with MTM are increasing and far exceeding the last published statistics on this disease. A harsh reality is that MTM takes too many lives, at too early an age. While we are incredibly hopeful for our futures, sadly families may face the unimaginable loss of losing a child while bravely battling this disease. Our community is here for you to help support you through this loss and to honor and remember your loved one. You are not alone. Those in our community who have lost a child to MTM find comfort in connecting with our community and allowing us the honor of helping to keep the legacy of your child strong and a valued part of our global MTM family. There are two ways to honor our precious children that have gone on before us; if you choose, we would love to honor any deceased loved ones affected by CNM/MTM on the joshuafrase.org [In Memoriam page](#). We would also encourage you to register their information on the [Family Registry for Centronuclear and Myotubular Myopathies](#) (Global Map); their information is just as crucial as your children who are still with us. To honor your loved one who has passed on the In Memoriam page, please email your child's photo along with dates, date of birth and date they received their angel wings to [info@joshuafrase.org](mailto:info@joshuafrase.org).

## **RESOURCES AND LINKS**

### **Joshua Frase Foundation**

To start with, you should begin by looking at the [Joshua Frase Foundation](#) website (<http://www.joshuafrase.org>). The JFF is the premier location for all things MTM. It has been funding research for over 17 years and has funded more than 50% of the research dollars worldwide. The Frase family has an amazing story and tons of materials that you will find helpful. Joshua Frase Foundation facebook page (<https://www.facebook.com/pages/Joshua-Frase-Foundation-supporting-Myotubular-Myopathy/132151230150159?ref=hl>) is also a resource for updates on research and happenings within our community.

### **Facebook Support Group**

There is also a wonderfully active and supportive community on facebook (<https://www.facebook.com/groups/MtmCnmFamilySupportandDiscussion/>). This is the forum where parents ask questions and discuss MTM topics.

### **Where There's A Will There's A Cure**

The family of William Whiston formed Will Cure (<http://www.will-cure.org/>) to raise funds to support MTM medical research. They help fund scientists and projects that are working toward new therapies for MTM children.

## **The MTM-CNM Family Conference**

There is a bi-annual MTM-CNM Family Conference (<http://www.mtm-cnm.com>) held in the United States. This is a grassroots event organized by a team of families and provides a wonderful opportunity to connect with other families and leading researchers in person. The conference website and facebook group (<https://www.facebook.com/groups/mtmcnfmfamilyconference>) are great for both resources and family networking.

## **Myotubular Trust**

Anne Lennox and Wendy Hughes, two parents of children affected by myotubular myopathy set up the Myotubular Trust (<http://www.myotubulartrust.com/>) in February 2006. They quickly became a leading source of funding and a leading source of information. Based in the U.K., they support worldwide efforts with a focus on European research.

## **Information Point**

The Information Point for Centronuclear and Myotubular Myopathy (<http://centronuclear.org.uk/>) was established in 2001 to provide information about centronuclear and myotubular myopathy, to provide support by bringing people with the condition together and to create awareness of this rare condition.

## **MEDICAL RESOURCES**

### **Physicians / Geneticists to Contact**

The Joshua Frase Foundation website provides a list of clinicians (<http://www.joshuafrase.org/resources/clinicians-to-contact.php>) who excel at providing care for children with MTM.

### **Shriner's Hospitals**

Shriner's Hospitals can be a wonderful resource for families with MTM. One needs to fill out an application found on the Shriners Website (<http://www.shrinershospitalsforchildren.org/>).

### **Muscular Dystrophy Association (MDA) Clinics**

The MDA ([www.mda.org](http://www.mda.org)) maintains a network of 200 specialized clinics across the United States and in Puerto Rico. Most MDA clinics are located in teaching hospitals, and many MDA clinic directors are university medical school professors as well as practicing physicians. MDA clinics are at the forefront of research and treatment methods; some clinics also serve as sites for clinical trials of the latest experimental therapies. MDA clinics utilize a multidisciplinary team approach, meaning individuals can see knowledgeable health care specialists from a variety of disciplines, all at one location.

### **MedicAlert Foundation**

Please consider using a medical alert identification. These are often worn as a bracelet or necklace and can alert first responders of critical medical issues in a medical emergency. (<http://www.medicalert.org/>)

**AUTHORS AND ACKNOWLEDGEMENTS:** The Joshua Frase Foundation would like to thank the following people for their time and effort in the production of this document: Carol Bowers, Sarah Foye, Betsy Grant, Scott Grant, Shannon Mashinchi, Erin Ward, Mark Ward, Daniel Whiston, Melanie Whiston, Marie Wood, Paul Frase and Alison Frase.

## GLOSSARY OF TERMS

**ACE inhibitors:** A group of medications that cardiologists use to help relax blood vessels and make it easier for the heart to circulate blood around the body. One of the main side effects of ACE inhibitors is possible lowering of blood pressure. Some common ACE inhibitors used in the United States are enalapril (Vasotec), lisinopril (Zestril), and perindopril (Aceon), but there are also many other options your child's cardiologist may choose. ACE inhibitors are also used to treat other conditions not related to the heart.

**Advocate:** To work with your child's health care providers as a team member to do the best thing for your child. This sometimes may mean speaking up when you are uncomfortable with a situation or plan of care, seeking out a second opinion, or identifying an advocate within your child's health care team.

**Anticoagulants:** A group of medications that thin the blood to prevent it from forming clots (which cause blockages in the blood vessels). The heart does not have to work as hard to pump thinner blood, but when people are on this type of medication they can bleed easily. Some common anticoagulant medications used in the United States are warfarin (Coumadin), heparin, and aspirin, but there are also other options your child's cardiologist may choose.

**Anticonvulsant therapy:** Medications that reduce the frequency and severity of seizures. Sometimes seizures can stop completely when a person is on an anticonvulsant. Some common anticonvulsants used in the United States are valproic acid (Depakote), levetiracetam (Keppra), zonisamide (Zonegran), and topiramate (Topamax), but there are also many other options your child's neurologist may choose. Anticonvulsants are sometimes used for other conditions not related to seizures or epilepsy.

**Arrhythmia:** A change in the rhythm of the heart's beating.

**Arthrogryposis:** A condition that results in an infant being born with multiple contractures. This can be an early symptom of CMD but may be confused with other causes for contractures seen at birth.

**Aspiration:** When something (food, liquid, mucus, etc.) goes into the lungs instead of into the stomach or out the mouth or nose. When a substance is aspirated into the lungs, it can lead to a lung infection (like pneumonia).

**Atelectasis:** collapse of part (or all) of a lung. This can be caused by a blockage of the airways or by pressure on the airways from outside the lungs.

**Bi-PAP:** Abbreviation for bilevel positive airway pressure, one of the most commonly used forms of noninvasive ventilation. Bi-PAP has two levels of airway pressure: a high pressure when the person breathes in and a low pressure when the person breathes out. A Bi-PAP machine can be programmed to cycle when the person breathes, or it can be set to a timed cycle.

**Blood gases:** A test to measure the concentrations of oxygen (O<sub>2</sub>) and carbon dioxide (CO<sub>2</sub>) in the blood, along with blood pH and bicarbonate level.

**Breath stacking techniques:** A type of respiratory therapy. The patient uses a special bag equipped with a one-way valve and mouthpiece to take a series of breaths without exhaling, expanding the lungs beyond what he or she can accomplish with a single breath. This stretches the lungs and opens clogged airways.

**Composite motor skills:** A way of measuring several different types of motor skills, such as handwriting and eating, and putting these measurements together to see how various aspects of a person's motor skills are doing overall.

**Continuous positive airway pressure (CPAP):** One of the most commonly used forms of noninvasive ventilation, CPAP increases the pressure of the air in the lungs for the whole time someone is using the machine. This is helpful for people whose airways get too small at times (as with obstructive sleep apnea), but CPAP is NOT recommended for individuals with breathing issues due to muscle weakness.

**Contracture:** A tightness in the muscles or tendons around a joint, which prevents the joint from moving the full amount. For example, a contracture in the knee can prevent the knee from straightening out or bending completely.

**Diagnosis:** The specific name of a medical disorder.

**Dysmotility:** When digested food does not move through the stomach or intestines at the right speed. Digested food moves through our body when the muscles in our intestines move like a wave to push the food along. Sometimes the wave moves too slowly and can cause constipation. Other times it moves too quickly and can cause diarrhea.

**Electroencephalogram (EEG):** A test of brain activity that looks for the cause of seizures by placing electrodes (monitors) on the head. The brain communicates to our body by sending messages (signals) from one nerve to another, producing a regular pattern we can expect to see when the brain is functioning normally. When an irregular pattern is seen on the EEG, a person can be at risk for having seizures, but the EEG doesn't tell us why that person may be at risk. To use an analogy, nerves are like telephone wires connecting houses. The EEG monitors the activity that is happening on the telephone wires but does not listen to the people talking to each other.

**Failure to thrive:** A term used to describe infants or young children who are not growing or gaining weight as expected. It is usually related to not taking in enough food to meet the child's calorie needs or to an inability to absorb nutrients from food.

**Fiber-endoscopic evaluation (or endoscopy):** A procedure that uses a thin, long tube and light to look inside a person. For example, endoscopy can be used to look at the intestines (colonoscopy) or lungs (bronchoscopy).

**Forced vital capacity (FVC):** The maximum amount of air someone can blow out after taking the biggest breath possible. The FVC can help measure if there is a problem with lung function, such as respiratory muscle weakness, or if an infection is present.

**Gas exchange:** The body process in which oxygen (O<sub>2</sub>) is moved from air to body tissues to use by the cells and carbon dioxide (CO<sub>2</sub>) is moved from tissues to the air. It occurs in the lungs and the bloodstream.

**Gastroesophageal Reflux Disease (GERD):** When stomach acid overflows out of the stomach and up into the esophagus (the tube that connects the throat to the stomach).

**Gastrostomy tube (G-tube):** A type of feeding tube that is surgically inserted through the skin and directly into the stomach. Some specific types of G-tubes are PEG tubes, Mic-Key buttons, and Bard buttons.

**Genetic mutation:** A change in a person's genes that alters something about his or her body or how it functions. Genes are the blueprints or directions for how everything in your body is made. We inherit genes from our biological parents. Our genes can be said to make up the letters that link together to produce sentences in an instruction manual. Using the same analogy, a mutation, when there is a bad change in the genes, is like a spelling mistake or when a sentence or section of the instruction manual is missing. Everyone has some changes in their genes, just as every book has some spelling mistakes. Most of these mutations do not cause major problems, but some genetic mutations can cause problems or diseases. For example, pretend that you bought a dresser and need to put it together at home. There may be some spelling mistakes in the instruction manual, but you can ignore them because you can figure out what to do. However, if words are missing from a sentence or a section is

missing from the instruction manual, you may not know that you have to use screws to hold all the parts of the dresser together. Or you may unknowingly leave the drawers in the box and turn the dresser into shelves.

**Gingival hyperplasia:** An overgrowth of the tissue that makes up the gums around the teeth in the mouth. This is often a side effect in patients who cannot close their mouths (because of hypotonia or muscle weakness) or in patients treated with phenytoin, a drug used to control seizures.

**Goniometry:** the measurement of a joint angle, or how much a joint can bend and extend.

**Holter monitor:** A device that is placed on a patient that allows an electrocardiogram to be produced over a longer period of time, usually 2 or 3 days. This device records the electrical activity of the heart and is used together with a patient diary to identify times of day or symptoms that may reflect a change in the recorded electrical activity. Once the Holter monitor is placed on a patient, the patient usually can go home and does not need to stay in the office or hospital. Heart muscle is different than skeletal muscle and not affected directly by MTM.

**Hypoplasia:** Underdevelopment of a body part. For example, midface hypoplasia is a flattening of the area around the nose that may be related to the use of a facemask.

**Hypotonia:** Tone is a term that describes the amount of tension or resistance to movement in a muscle. Hypotonia refers to low tone (also sometimes called *floppy*), and the body part moves easier than it should. High tone is called *hypertonia* or *spasticity* and is when a joint is stiff. Tone is a different measure than strength (a hypotonic child can have residual strength in muscles), but it is often hard to tell the difference between tone and strength in infants.

**Insufflator-exsufflator device:** A machine used to help encourage good lung function by simulating a cough; the lungs are filled full of air (like taking a deep breath) and then the air is briefly sucked out of the lungs (like a forceful cough). Usually, these devices are set on a cycle of a certain number of coughs each time it is used. These machines are also called *coughalators* or are known by the brand name CoughAssist. Some children say that these machines take some time to get used to using, but that once that happens they feel much better after using it.

**Intrapulmonary percussive ventilation (IPV):** A type of chest physical therapy in which a device (machine) provides very fast vibration to the chest to help mobilize secretions (get mucus out of the lungs). There are many different types of IPV devices; some are hand-held; another is a vest that is worn by the patient.

**Jejunostomy tube (J-tube):** A type of feeding tube that is surgically inserted through the skin and directly into the lowest part of the stomach, a region called the jejunum. This type of tube decreases the risk of reflux.

**Magnetic resonance imaging (MRI):** A detailed picture of the structure of a body part. An MRI provides much more details (that is, has a higher resolution) than a CT scan or X-ray; in addition, an MRI does not use any type of radiation. An MRI is useful when looking at soft tissues like brain and muscle, but is not the ideal tool for looking at bones. To use an analogy, an MRI of the brain is like looking at the “Satellite” view on Google Maps. It can tell us where the houses, streets, and parks are and what they all look like or if a tornado came through and caused lots of structural damage. But with an MRI (the picture of the city) we cannot see the individual brain cells or nerves (people in the houses), nor determine the function of the brain (tell if the people are going to work on time or if there is a lot of traffic one day). An MRI can only show us structure

**Malignant hyperthermia:** An allergic reaction to some types of anesthesia (medicines given to people to make them sleep through a procedure). This can be a life-threatening reaction that causes the body to become too hot. Like all allergies, only some people have this problem, but certain genetic mutations may increase the risk of malignant hyperthermia. Please refer to this [anesthesia guideline](#) for respiratory management for those preparing for surgery; also share with your physician.

**Multidisciplinary care:** When health care providers with different areas of expertise work together as a team—for example, when a neurologist, pulmonologist, physical therapist, and dietitian all work together to help improve the health of one patient.

**Multisystemic:** When multiple different body systems are affected by a disease or condition or when they are monitored or examined together by a health care provider.

**Myometry:** The formal name for measuring muscle strength using a special device that gauges the amount of force exerted by a given muscle or muscle group.

**Nasogastric tube (NG tube):** A type of temporary feeding tube that is inserted through the nose and ends in the stomach.

**Nissen fundoplication:** A “knot” that is surgically tied in the upper part of the stomach to help prevent severe gastroesophageal reflux disease (GERD).

**Noninvasive ventilation:** A way to help people who cannot breath on their own or are not breathing well. This type of ventilatory (breathing) support is given by noninvasive methods, like through a mask rather than through an invasive method like a tracheostomy tube, and can be used at specific times, like only at night or only during illnesses. Noninvasive ventilation is often preferred over invasive ventilation. Positive airway pressure (PAP) is an example of a noninvasive ventilation technique.

**Orthosis:** An artificial or mechanical aid, such as a brace, to support or assist movement of a body part. Examples of orthoses include AFO, which stands for ankle-foot orthosis. An AFO is a one-piece hard plastic splint that is molded to the posterior lower leg and under the foot, is typically fastened with Velcro, and can be worn over a sock and in a shoe. The AFO provides support to children with low tone and may assist them with achieving and maintaining ambulation.

**Oximetry:** Measurement of the oxygen content of blood.

**Palliative care:** A type of multidisciplinary care for people with serious medical diseases. Palliative care is different from end-of-life care or hospice care. The goal of palliative care is to improve the patient’s and family’s quality of life by reducing the symptoms of the disease.

**Peak cough flow:** A measurement of how hard someone can cough; this helps measure lung function and the person’s ability to clear secretions (that is, get mucus out of the lungs).

**Polysomnography (sleep study):** A recording of the many changes in a person’s body that happen during sleep. During the study, a sleeping patient’s lungs, heart, and brain function, along with eye movement and muscle movement, are monitored using different tests. It is useful in understanding the cause of daytime fatigue.

**Positive airway pressure (PAP):** A type of noninvasive ventilation that was originally developed for people with sleep apnea but is also used for people with neuromuscular diseases. There are two types of PAP: continuous positive airway pressure (CPAP) and bilevel positive airway pressure (Bi-PAP). Once the airway is open with this type of machine, a person can breathe normally.

**Prognosis:** How a disease is expected to change over time and what those changes mean for your child’s health and life.

**Progression:** The process or path that a disease takes over time.

**Psychometric testing:** The name for a group of tests that evaluate learning, cognition, behavior, mood, and personality traits. This type of testing can also be called a *psychoeducational evaluation*. The specific tests performed are not the same for every child. They can change due to a child’s age or specific concerns that need to be evaluated.

**Pulmonary function tests (PFTs):** A group of tests that measure how well the lungs work to take in and release air and how well they move oxygen into the bloodstream.

**Scoliosis:** An abnormal sideways curve in the spine (back bones) that makes the spine take on a “C” or “S” shape. This type of curve is different than a curve in the lower back (lumbar region) which makes the stomach stick out (lumbar lordosis) or a curve in the upper back (thoracic region), which some people call “hunchback” (thoracic kyphosis). When both kyphosis and scoliosis are present, this is called kyphoscoliosis.

**Seizure:** An excessive surge of electrical activity in the brain. This surge can stay in only one part of the brain (a partial/focal seizure) or surge through the whole brain all at once (generalized seizure). Because the brain controls everything we do, a seizure can look different in different people, depending on where the seizure is coming from in the brain. Some people can have seizures where their whole body shakes, or just an arm or leg shake. Other people can have a seizure that just looks like they are staring, or staring can be combined with abnormal movements of their mouth, eyes, or hands. The definition of epilepsy is two or more unprovoked seizures. If you have concerns about potential seizure activity in your child, please speak with your health care provider. For more information on seizures and epilepsy, please see [efa.org](http://efa.org) or [epilepsy.com](http://epilepsy.com).

**Sleep apnea:** Abnormal pauses in breathing during sleep. It is normal for the respiratory (breathing) rate to slow down when someone is sleeping; however, sometimes it slows down too much. If someone has long pauses between breaths, carbon dioxide can build up in the bloodstream. When this happens, it is possible that not enough oxygen gets to the brain (hypoventilation). Sleep apnea is stressful for the body. When someone has untreated chronic (long-term) hypoventilation, it can lead to heart failure or other multisystemic problems.

**Spirometry:** The most common of the pulmonary function tests, spirometry measures the amount of air entering and leaving the lungs.

**Subluxation:** When a bone comes partially out of a joint but does not completely dislocate. In CMD, the hips frequently subluxate.

**Torticollis:** A type of neck contracture in which the neck is twisted, making the head tilt to one side and the ear move closer to the shoulder. When a child has torticollis, they cannot turn their head all the way from one side to the other.

**Ulnar length:** The length of the lower part of the arm, from the wrist to the elbow, which can be used to calculate height when someone cannot stand up straight.

**Valproic acid (VPA):** One of the specific types of anticonvulsants. This medication is known by the brand names Depakote (pills/capsules) and Depakene (liquid).

**Videofluoroscopy:** A type of X-ray that takes a video while someone swallows foods or liquids to test for aspiration. This test is also called a *modified barium swallow study*.