

A FAMILY GUIDE TO MYOTUBULAR MYOPATHY

 Joshua Frase
FOUNDATION™



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TABLE OF CONTENTS

	About MTM.....	4
	Respiratory Overview.....	12
	Home Equipment.....	20
	Feeding and Gastrointestinal Care	28
	Orthopedic Care.....	30
	Mobility	32
	Other Issues and Considerations	34
	Medications	38
	Caring for Your Child at Home	40
	Insurance and Medicaid	46
	Research, Treatment and Registries	47
	Honoring & Remembering	48
	Resources & Links	50
	Glossary of Medical Terms	54
	Durable Medical Equipment (DME)	64
	Transportation, Car Seats, Strollers & More.....	68
	Contributors	78

DISCLAIMER: The information and advice published or made available in this booklet is not intended to replace the services of a physician. This booklet has been written and edited by a number of experts with experience in MTM. It is suggested that parents use this booklet as a reference, and share it with their child's clinician. Any action on your part in response to the information provided in this booklet is at your own discretion.



A STARTING POINT FOR THOSE NEW TO THE MTM COMMUNITY.

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 treatments 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 diagnosis of Myotubular Myopathy (MTM, or, X-linked myotubular myopathy, XLMTM) can be scary and overwhelming. We hope you take comfort in the fact that others have experienced what you are going through, and we are eager to share our collective knowledge with you to ease you through the first steps on this journey. The next few pages will contain vital information to take you from birth to caring for your child at home, and beyond...



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. The guide was created in 2013 and updated in 2020 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 doctor's fault, MTM is a rare orphan muscle disorder that few have heard of and many doctors may never see a single case their entire career. 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, in 2012 there was an 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.

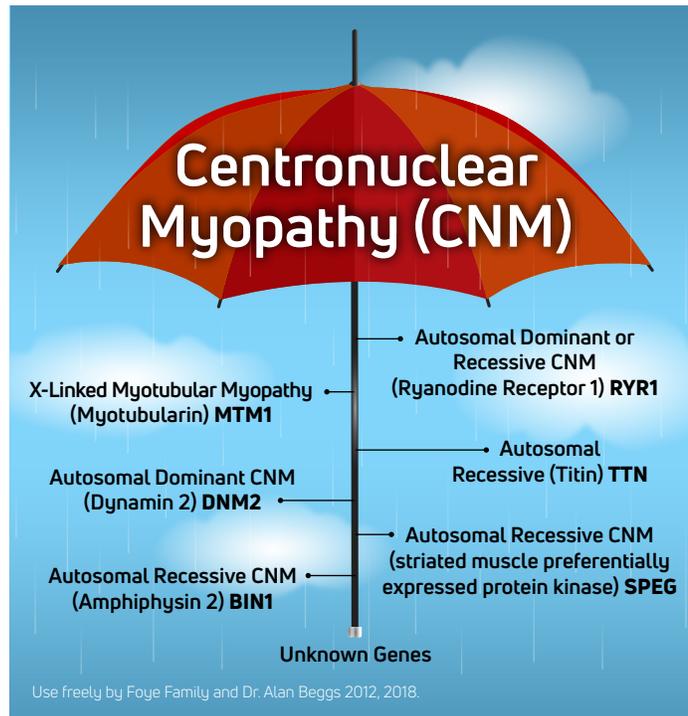
If you are reading this you probably have a child that was born “floppy”. Your doctor may have told you that your child may have a myopathy and you scoured the internet for myopathy information. Your child may have had a muscle biopsy and you have been told that your child has Centronuclear Myopathy 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 vs 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 centronuclear myopathies as shown in the umbrella diagram, or, may have a different muscle disease altogether such as congenital myotonic dystrophy. Genetic testing is also important because it is often needed to enter into clinical trial.



Use freely by Foye Family and Dr. Alan Beggs 2012, 2018.

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 and often the lungs and diaphragm. 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.

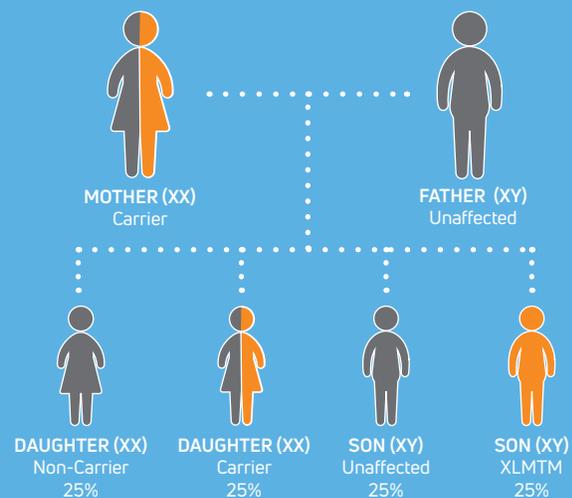


Genetics and Diagnosis

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, though an increasing number of affected girls and women are being identified. Sometimes a boy is born with MTM due to a spontaneous change in the gene, meaning the mother is not a carrier (de novo mutation). This occurs about 10% of the time. 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](#), or [Jim Dowling's laboratory](#), (also see references below) to discuss getting genetic confirmation.

XLMTM is an X-Linked condition, caused by a change in the *MTM1* gene on the X chromosome.

- Females have two X chromosomes and males have one X and one Y
- Females who have a genetic change in *MTM1* gene are healthy and are known as **carriers**
- Males who inherit a genetic change in the *MTM1* gene from their carrier mothers have **XLMTM**



ABOUT MTM

The Joshua Frase Foundation



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 the best way to treat 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. If a Special Needs Palliative Primary Care physician is available, they can be wonderful to help manage all the information coming in from specialists.

A good resource for physician recommendations is other parents. They have invaluable experience and advice concerning the care of MTM children. Frequently used Facebook groups "[MTM/CNM Family Support & Discussion](#)" and "[CNM/MTM Families United](#)" are meeting places 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. Additionally, there has been a family conference every two years where families gather and listen to professionals and meet other CNM/MTM affected families here in the US and in Europe.

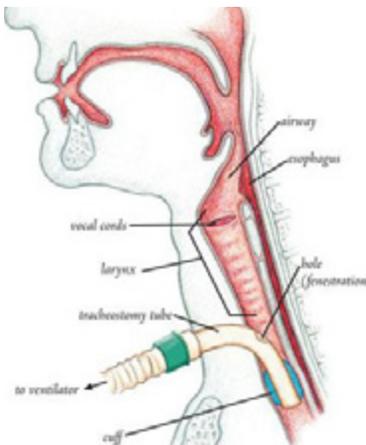


RESPIRATORY OVERVIEW

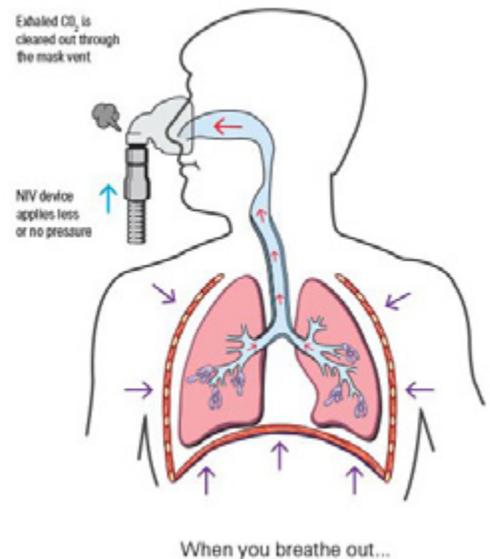
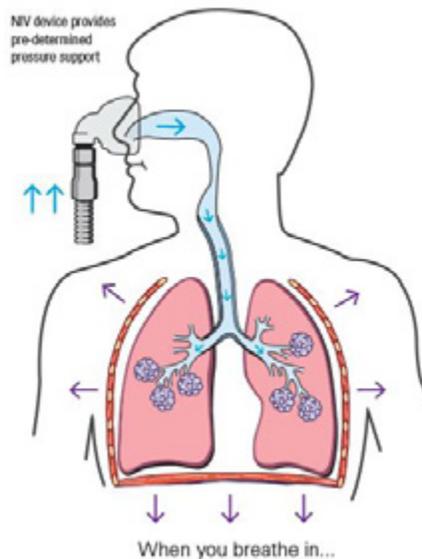
All MTM children have weakened muscles which makes the act of breathing difficult and/or impossible. There are many muscles that help us to breathe. The diaphragm is a large muscle at the base of the lungs; when it does not move well due to weakness, our ability to take a breath is greatly affected. With a weakened diaphragm and other weakened muscles the ability to breathe will be reduced (hypoventilation) and normal gas exchange of oxygen and carbon dioxide cannot happen. When there is improper gas exchange there will be a decrease in oxygen (hypoxemia) and a buildup of carbon dioxide (hypercarbia). If this goes untreated, an emergency situation arises, and without immediate intervention death will occur. This is the reason MTM children must have excellent respiratory support. Other respiratory management issues include helping your child manage oral secretions and assisting with coughing. These will be explained in a separate section below but they are very important in maintaining good respiratory care.

MANAGING YOUR CHILD'S BREATHING IS THE NUMBER ONE PRIORITY FOR MOST MTM CHILDREN

Invasive Support



Non-Invasive Support



Finding the Right Doctor / Pulmonologist

Early on you will most likely meet with a pulmonologist who will work to manage your child's respiratory needs. A pulmonologist is an Internist who has done an additional 3 years of study to specialize in lung diseases such as Asthma or Cystic Fibrosis. It is important to understand that MTM is not a lung disease and should not be treated that way. However, as discussed above, weakened muscles cause life threatening problems and having a pulmonologist who is familiar with this issue is imperative. Some of the treatments are similar to having a lung disease but some treatments are very different.

Treatment Options for Breathing Problems

Approximately 90% of MTM children require breathing support at birth, and the majority are placed on a ventilator (breathing machine) due to their inability to breathe adequately. The breathing tube that is used at that time cannot remain in place for an extended time due to complications that it will create. The hospital team's goal will be to try and remove the tube in the hope that your child will be able to breathe on his own. They will lessen the amount of support the ventilator provides to see how well your child will do. Most MTM kids will not be able to do this, due to severe weakness and the hospital team will talk to you about performing a tracheostomy. Between 60-80% of MTM boys will ultimately utilize a tracheostomy for support. Those that do not require tracheostomy will often need support through some other form of mechanical support such as BiPaP or a negative pressure ventilator.

We estimate that 15-30% of MTM kids are managed with non-invasive ventilation, and that this number may be increasing with advances in respiratory care. We will explore these options here.

RECENSUS

In 2018 a natural history study called **RECENSUS** was published. The goal of **RECENSUS** was to define the disease burden and medical need of children with MTM.

RECENSUS collected and analyzed data on things such as respiratory support, frequencies of hospitalizations and surgeries and other complications from 112 MTM patients across six different clinical sites. With the collaboration of the scientists and families that participated, we were able to get a clearer picture of how MTM presents and is managed over time and across our patient population.

This knowledge can help families inform themselves of the typical disease process and presentation to better inform their care decisions. We suggest forwarding a copy of **RECENSUS** to your child's physician for advance review. That way you can bring a copy with you to an appointment and discuss its findings and implications to your child's individual care plan.

The **RECENSUS** study is a great example of how participation in a natural history research study can help drive understanding of the disease for researchers and our community. A larger updated **RECENSUS** study was released in 2019. Review the results, [click here](#).



112
MTM PATIENTS

+



6
CLINICAL SITES

+



100%
COLLABORATION OF
FAMILIES + SCIENTISTS

See link under *Research Treatments & Registries Section* to read **RECENSUS**.

The 2019 **RECENSUS** is an international, retrospective study of mortality and respiratory support in patients with XLMTM. This is a great example of how participation in a retrospective analysis can help drive understanding of the disease for researchers and our community. You can review the article here [here](#).

Tracheostomy and Ventilators

The majority of kids with MTM have such severe muscle weakness that long-term ventilation via a breathing machine is the only answer for their survival. In order to have that, a tracheostomy must be performed. A tracheostomy is a hole (ostomy) in the front of the neck with another hole in the trachea (windpipe) where a tracheostomy tube or trach tube will be placed. A ventilator can then be attached to the outside of the trach tube and the settings will mimic the act of breathing for your child.

The trach tube also provides access for removal of secretions. Making the decision to allow the doctor to perform a trach on your child is often a difficult one. It is important to understand that many children thrive once they are getting adequate breathing support because they no longer have to work so hard to simply breathe. Their energy can be used for other things such as playing and interacting with loved ones. Understanding the whole scope of what life will look like for you and your child will be important. Being prepared and having an idea of what to expect will help with the adjustments that your family will need to make.

ADVANTAGES OF A TRACHEOSTOMY

Your child can have round-the-clock life-sustaining breathing support

They will likely have more energy since he no longer has to work so hard to breathe

Direct access to airways for suctioning purposes and for nebulizer medication delivery

No obstruction of the face

DISADVANTAGES OF A TRACHEOSTOMY

It will require newly learned skills and the care is more complicated

A properly trained caregiver will always need to be with your child

Some will have difficulty with speech and swallowing

Coughing to clear the smaller airways is difficult

There is a risk of infection at the trach site (stoma)

Once, your child has received his tracheostomy (if needed), the hospital team will attach a ventilator (breathing machine) to it and use settings that are specific for your child's needs. They will start teaching you about the ventilator, how to care for your child's trach, how to change his trach, what to do in an emergency and all the other care that is needed. Having a good social worker on your team can also be very helpful when planning your child's discharge home. It will likely feel very overwhelming but trust that you will be able to learn and become an expert in your child's care. The ventilators that are used in the hospital are quite large but home ventilators are relatively portable, allowing kids to enjoy school, travel and have a full range of experiences. It will take extra planning and preparation but over time you will see that a full life can be achieved.

Non-Invasive Support

As previously mentioned, there is a small group of MTM children who do not need as much breathing support. They not only have the ability to breathe on their own but some can, eventually sit up and learn to walk. Even among this group there are different levels of support needed. This is why having a team of professionals who can properly evaluate your child based on his abilities and goals is valuable. It is common among this group that during any type of illness, their ability to breathe without support can be severely compromised. Being able to recognize the increased need for support and provide the interventions is very important.

The most common form of support is called BiPaP (BiLevel Positive Airway Pressure). CPAP or Continuous Positive Airway Pressure should never be used in patients with a neuromuscular disease such as MTM. BiPap works by delivering airway support via a mask which either fits over the nose or over the nose and mouth. With young children a mask that covers the nose and mouth is often not recommended however. The mask will be attached to either a BiPap machine or a small portable ventilator. The settings will be determined by your child's physician and should be reevaluated at least yearly. Whenever your child is sick your doctor may want to consider a temporary setting change if he is having problems breathing. Often, kids with a milder form of MTM will use the BiPap when sleeping to prevent hypoventilation and during an illness. Some kids will need to use BiPap for longer periods of time during the day. Again, because some of our kids are stronger than others, a professional will be able to help guide you.

Another form of non-invasive support is via a negative pressure ventilator (NPV). This works like the old iron lung that people were placed in when polio was a huge epidemic. Just like the ventilator has become smaller and more portable so has the NPV. There is a cuirass or shell that fits over the chest and trunk area, it attaches to a ventilator which forces air in and out of the lungs by using negative pressure. As your child takes a breath the ventilator will sense this and expand the lungs more thus decreasing the workload of your child's muscles. It mimics the way we breathe naturally which is by using negative pressure.

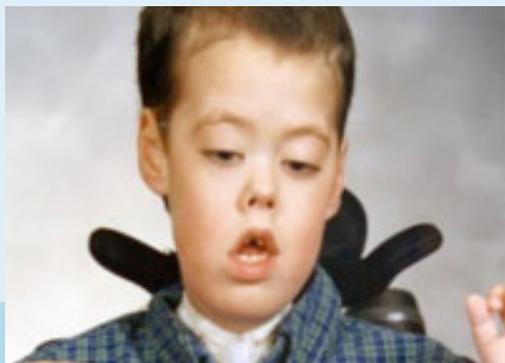
Knowing about and considering all the options based on your child's abilities will help you and your team of experts develop a plan of care that is right for your child and your family.

Although the delivery of breathing support may differ, there are other pieces of medical equipment that most all MTM kids use. You may hear the terms "pulmonary hygiene" or "pulmonary toilet", which basically refers to the acts of keeping the lungs healthy and functioning. All the machines listed here make up what is needed. These help to keep your child healthy and to provide care when ill. They are vital in the daily ongoing care of your child. Even though you may not be using them now there will be a time that you will and to have them already in the home is important.

ADVANTAGES OF BIPAP AND/OR NPV	DISADVANTAGES OF BIPAP AND/OR NPV
No surgery needed	Skin irritation from mask or cuirass
Ability to talk and swallow remain intact	Mask interferes with communication and interactions
May use only when needed	May have stomach distention
Diaphragm muscle continues to work	Mask/cuirass may not be tolerated by a young child

RESPIRATORY OVERVIEW

The Joshua Frase Foundation







HOME EQUIPMENT

Ventilators

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. A speech therapist can help your child to learn how to speak with a trach in place. This often involved using a speaking valve placed on the tracheostomy tube which allows air into the patient from the ventilator, but forces the air out past the tracheostomy tube and through the vocal cords and out the nose or mouth. Some individuals with MTM also communicate with sign language or assistive communication devices.



Respironics Trilogy

BiPAP (BiLevel Positive Airway Pressure) Support

BiPap is a term coined by a company who first developed a machine for this type of therapy. The machine and its abilities differ greatly from the well known CPAP machines in that the machine adjust the pressure to a lesser setting when the patient exhales. This allows for a more natural and much easier exhalation. There are newer machines now that are even more sophisticated and help to support breathing without the need for a tracheostomy. A popular machine that is used for a lot of patients with weakened muscles is a Trilogy ventilator. It is basically a very sophisticated "BiPap" machine. There will be two primary settings that your child's doctor will set based on your child's needs. They are known as the Inspiratory Positive Airway Pressure or IPAP and the Expiratory Positive Airway Pressure or EPAP. The EPAP setting will be less than the IPAP setting allowing for your child to exhale against less airway pressure. The more sophisticated machines such as the Trilogy have an AVAPS (Average Volume Assured Pressure Support) setting. The machine will automatically adjust to changes in order to maintain an adequate tidal volume. Tidal volume is defined as the volume of air that is inhaled or exhaled in a single breath. This can be helpful during times when your child's needs may change such as during a respiratory illness.

CPAP

Continuous Positive Airway Pressure (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 should **never be used** as a way to support breathing in patients with neuromuscular disease and trouble breathing.

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 to remove mucus from airways and saliva from the mouth and nose. Muscle weakness from MTM affects swallowing and therefore the suction machine is also used to remove saliva and mucus that accumulate in the mouth, nose and lungs. 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.



DeVilbiss 7305

Mucus plugs can be fatal as they can block the 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, a pulse oximeter, nebulizers, humidifier for humidity, suction catheters, and chest physical therapy can help you monitor and maintain your child’s daily pulmonary hygiene.



IMPORTANT NOTE: If you are leaving the hospital without the following equipment inquire why that is and then ask your doctor to provide you with a prescription for the following equipment listed on the following pages.

Pulse Oximeter

Commonly used names are pulse ox or sat (saturation) monitor. This machine is a must for all MTM kids. This monitors the oxygen level or oxygen saturation in your blood. It will also detect your heart rate. It will alarm whenever your child's oxygen levels drop and/or when the heart rate detected is below or above normal. Check with your doctor to learn the correct O₂ (oxygen) parameters for your area.



Masimo Rad-8 Pulse Oximeter

Depending on the elevation of where you live, the alarm will be set to go off when approaching the danger zone. If the low oxygen alarm goes off this is an emergency situation. You will learn the different things to check for such as a possible disconnect from ventilator support, a plugged trach tube, or the low reading may be the beginnings of a respiratory infection. The daily continuous monitoring of your child's oxygen levels are a must. Getting to know your child's normal heart rate at rest can also help to signal you when trouble is brewing.

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. Check with your doctor but an aggressive airway maintenance program will improve the survival rate of your child.

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 (or wrap for very small children), 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. However, neither CPT or a percussive vest can be the only airway clearance in a patient with neuromuscular weakness and poor cough because they only free up mucus and make it easier to clear with coughing.

Intrapulmonary Percussive Ventilation (IPV)

Another device that can be used to loosen secretions prior to removal with the cough assist device is an IPV. Depending on the child, it may aid in better mobilization of secretions compared to the CPT vest. Some children respond better to an IPV and some to vest therapy both in tolerance and efficacy. Take advantage of your time in the hospital to trial both and see which works best for yours. This machine works by delivering rapid but small bursts of air directly into the airway through either an adapter for the ventilator circuit or a face mask.

This causes the airways to open up and the secretions to separate from the pulmonary lining so that they can be more easily removed. Medications can be nebulized with the machine while it is running to help loosen secretions, as well. Research has shown that depending on the set up, not as much of the medication may be delivered to the patient using the device, so keep that in mind when choosing the order of your therapies. Saline can be used while it runs as well. In some hospitals, they refer to the IPV as the "Metaneb". This is a version of the IPV made by Hill-Rom. It is not adaptable to the home environment but is very similar in its action to the one you would be able to get at home.

Cough Assist

The cough assist, also known as an insufflator-exsufflator, also helps move the mucus up and out and is the only effective way to replace a cough in a patient with neuromuscular weakness. Because of this, it is an essential part of your child's respiratory care on a day to day basis when your child is well and most importantly when your child gets sick.



Phillips Respironics T70
Cough Assist

The cough assist is a machine that mimics a cough. The machine connects to your child's tracheostomy tube if they are 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 can be used in conjunction with the other treatments described above, but is often effective as the only airway clearance device.

It is important to remember that when your child is sick he or she will need frequent cough assist treatments, just like how often you need to cough when you get sick. This may mean using the cough assist every 1-2 hours when your child is sick, and occasionally more frequently as needed. This is very important if your child is in the hospital; he/she will need an order for the cough assist that specifies how frequent, and at what pressures, the treatment can be given. Your child should have an as needed or "PRN" order for the cough assist so that he/she can use it as frequently as needed.

In some cases a nebulizer treatment of Albuterol or saline can be used to help free up mucus, 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 secretions that were loosened by the first two treatments.

If your child does not have a cough assist you should ask your pulmonologist if your child may benefit from one. Your insurance company may try to deny you either your cough assist device or your secretion mobilization device (cough assist/IPV) and only cover one. Remind your physician that in myopathies the patient not only needs help with secretion mobilization, but also with secretion removal (cough). This will help guide them with writing the medical necessity letter. Many studies support the cough assist device as the standard of care for myopathic patients.



ALARM FAILURE AND FATIGUE 101

Alarm fatigue is a dangerous thing, even in hospitals, where fatalities and injuries due to this phenomenon do occur.

What Is Alarm Fatigue?

When your alarms are such that they go off all the time for non-emergent reasons and you become numb to them. They thus lose their power to alert you to an emergency when it happens.

What Is Alarm Failure?

Alarm failure is when a piece of equipment either delays to or fails to alarm in an emergency. There are various reasons this can happen. [See this document on the JFF website for more info.](#) Remember that alarms are only useful if they are set appropriate to your child, and are plugged in or adequately charged. Work with your pulmonary team to tailor your equipment to your child and always make sure to have redundancies in case of machine failure.



As parents of ventilator dependent children, understanding the factors that contribute to alarm failure and fatigue is so important to avoid unnecessary hypoxic injury and loss of life. Every second matters when dealing with emergent respiratory events; as we say in the field, “time is tissue”.

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 and 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 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. Supplemental oxygen should never be given without first asking why and trying to treat the reason. The most common reason is a mucus plug in the lungs and the treatment would be using the cough assist to clear the mucus.



Oxygen Tanks

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 a cough assist treatment and suctioning a mucus plug, or treating for pneumonia. Do not give or increase oxygen without also attempting to address the underlying problem and seek advice from your doctor if your child needs oxygen. Oxygen can often mask ineffective ventilation allowing CO₂ (carbon dioxide) levels to build in the blood and depress the drive to breathe. [Most Common Respiratory Management Mistakes.](#)

Resuscitation “Ambu” Bag

An Ambu Bag (a form of modified manual resuscitator) 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. If your child requires oxygen, an Ambu Bag can easily be connected to your oxygen source. In the event of an emergency an Ambu Bag can be used to get deep breaths.



Ambu Bag



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 feeding 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 breathing and swallowing. Your doctor may prescribe a particular formula or you may decide to make a homemade formula, using a special blender like a Vita Mix. A Vitamix is a powerful blender which liquefies food so it can pass through a small tube. Our community has had great success with the Vitamix 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 dietician to insure adequate nutrient and caloric intake.

If you chose to use formula, these are questions you should ask your child's doctor when determining what formula is best for assimilation and absorption.

- 1 – How important are amino based formulas?
- 2 – Are there formula's more beneficial for liver function?
- 3 – Are amino acid based formula's less likely to raise liver enzymes?

Constipation

Constipation, difficult bowel movements or having 3 or less stools in a week affects 50% of children in the MTM Community and can be uncomfortable. Muscle weakness, decreased activity, diet and medications can contribute to constipation and slower motility of stool through the bowel. Stool moving slowly through the bowel can become hard, dry and difficult to pass, which can cause the abdomen to become distended. Proper management can help alleviate this problem.

Daily routine, toilet training, diet, proper hydration, natural remedies and medications like stool softeners, laxatives, prune juice, suppositories, and enemas can be helpful. Consult your physician before starting a bowel routine to choose the most appropriate solution for your child.

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 (gastrostomy tube) is a small surgically placed device, commonly called a “button”, that enters directly into the stomach, and is a more permanent option than the NG tube. It is used to allow liquid nutrition to be given directly to the stomach without the need for the person to swallow.



G-Button

Swallowing requires many muscles working in a coordinated fashion, including the epiglottis which is 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. Vomiting can cause irritation of the esophagus, but also there is a risk for aspiration. Aspiration is when secretions, vomit or liquids are breathed into the airway. The muscles that limit reflux are also weak in MTM children. Children 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.



AFO's

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. As your child grows, the scoliosis may become an issue for comfort and breathing. Surgery to correct the curve may be considered. It is a major procedure, but can offer relief.



TLSO Brace

Another common orthopedic issue common to MTM children is hip subluxation. This is when the ball of the femur partially dislocates from its socket and is due in part to both the hypermobility of the joint and the absence of weight bearing. Working with your therapy team to acquire a stander to assist your child in weight bearing exercise to maintain joint stability can help with this issue as well as give proper input into the bones to prevent further weakness and complete dislocation or fracture. The stander secures your child in an upright position at progressively challenging angles while securing the knees, hips, and chest and has a head rest. A device for simulated crawling called the creepster crawler uses a similar concept for weight bearing into the knees and arms.

Keep in mind that procuring specialty equipment from insurance can take 6 months or longer so don't delay in getting started. Be aware of your other options as well such as MDA equipment closets that lend equipment to families when they have it available as well as other families on the Facebook support groups that may have equipment they are no longer using. Please visit [JFF's Equipment Sharing Program](#) for this purpose.



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 joystick 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.

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



MOBILITY

The Joshua Frase Foundation





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 hepatis (“peliosis of the liver”). It is not clear if or how this may be linked to the MTM1 gene which mainly affects muscles, but there are still many unknowns 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. However, there is no good evidence that screening with ultrasound or routine liver function blood studies is able to detect hepatic peliosis prior to life threatening hemorrhage. If a child with MTM experiences unexplained fever and abdominal pain, it is important to consider peliosis as a cause, and, at that moment, imaging (ultrasound/CT/MRI) should be suitable for detection.

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. In some cases, medication is needed to help resolve abnormal liver function.

Seizures

Some children with MTM have also experienced seizures, though the prevalence of this is rare (4% in one study). 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, partial seizures, and tonic clonic seizures. The cause of the seizures in these children is not well understood, though sometimes they can result from prolonged lack of oxygen, fever, 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 non verbal and weak, and can have extended blinking times. Absence seizures typically last seconds to several minutes. Tonic Clonic seizures are more obvious with the stiffening and jerking of the body. This type can last several minutes to much longer. Seizures can present differently, from absence staring off type, a small twitch or repetitive movement in a limb or face, to more significant tonic clonic body jerking type.

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 of time. 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 during the seizure.

An EEG (electroencephalogram) test can help establish the potential risk of seizures, and rarely will capture a seizure itself. As such, an EEG will only show current brain activity, it will not show if past seizures have occurred; it is thus most useful for understanding the risk for seizures, and for guiding choice of seizure medicines if they are needed. In some cases, a prolonged (up to 24 hours or longer) EEG may be helpful in catching a recording of a seizure as it’s happening and this may help your neurologist determine whether “spells” your child is experiencing are actually seizures, or are instead other types of movements or behaviors. 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, undescended testicles 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

Children with CNM/MTM tend to blink less frequently and less fully. This can lead to dry eyes and tearing, and sensitivity to bright light. Your child may not fully close his eyes while sleeping, which also leads to dryness. Signs include redness and tearing. Long-term, corneal abrasions can occur and scarring of the surface of the eyes.

Several interventions have helped others protect their eyes. Lubricating drops applied frequently during the day, and an OTC (over the counter, which means non-prescription) ointment at night are helpful. Preservative free ointments and drops are recommended. There are some night time goggles that have been found to be very protective and help sustain continuous moisture. There is a surgical intervention which may help, but carries risks of its own, where a stitch is placed to hold the lids closer together.

The goggles are very supportive because of the moisture seal, and keeping the eyes closed. Here is a link to a page showing an example of eye ointment that can usually be bought at a local pharmacy: <http://www.refreshbrand.com/Products/refresh-lacrilube>

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 be painful and become infected, requiring antibiotic treatment. Some children have small ear tubes (also called myringotomy 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, and should be considered in all individuals with MTM. It is also important to optimize sun exposure if possible, as this is the best source for vitamin D for the body. Some families have tried biophosphonate treatments to help strengthen the bones.

Other Complications

There are other complications that have been documented and/or self-reported and shared within our community. These complications (reported in less than 10% of MTM individuals, and often much less than that) 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, abnormal heart rhythms, 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), HIE (hypoxic ischemic encephalopathy, brain damage caused by lack of oxygen) 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. Neuromuscular blocking agents (such as succinylcholine) should be avoided. Conversely, individuals with MTM are NOT at risk of developing malignant hyperthermia (MH).



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.



MEDICATIONS OFTEN USED BY MTM CHILDREN

Your child may be prescribed medications of one kind or another. Each child is unique and has their own list of medications prescribed for them.

The following is a list of some of the medications used:

ALBUTEROL IAPRTROPIUM BROMIDE AND ALBUTEROL SULFATE (DUONEB®) - used to prevent and treat wheezing and shortness of breath caused by breathing problems.

EYE DROPS / EYE OINTMENT - helps keep eyes moist, and helps with redness, itching and watery-eyes.

FLUTICASONE PROPIONATE (FLOVENT®) - Inhaled steroids, also called corticosteroids, reduce inflammation in the lungs. They're used to treat asthma and other respiratory conditions. Inhaled steroids reduce inflammation in the lungs, allowing you to breathe better. In some cases, they also reduce the production of mucus.

PAMIDRONATE - used to treat high blood calcium levels and certain bone problems (bone metastases/lesions). It is also used to treat a certain type of bone disease (Paget's disease) that causes abnormal and weak bones.

PHENOBARBITAL - commonly used to treat seizures in young children; may be administered intravenously, injected into a muscle, or taken by mouth.

PREDNISONE - Prednisone provides relief for inflamed areas of the body. Prednisone is a corticosteroid (cortisone-like medicine or steroid). It works on the immune system to help relieve swelling, redness, itching, and allergic reactions.

PREVACID® - used to treat and prevent stomach and intestinal ulcers.

PULMICORT® - used to control and prevent symptoms (wheezing and shortness of breath) caused by asthma. It works directly in the lungs to make breathing easier by reducing the irritation and swelling of the airways.

PULMOZYME - used to improve breathing and reduce the risk of lung infections. It is used along with other treatments (e.g., chest physical therapy, medications, nutritional supplements). It works in the airways by decreasing stickiness/thickness of mucus so that it can be cleared from the lungs more easily.

SINGULAIR® - a medication commonly use to prevent asthma attacks and allergy symptoms.

TRIAMCINOLONE - Triamcinolone Acetonide Ointment, (brand names: Cinolar, Kenalog, Triderm) is a topica corticosteroid ointment for the skin, used to treat inflammation or irritation caused by conditions such as allergic reactions, eczema, and psoriasis.

URSODIOL - used to dissolve certain types of gallstones, to prevent gallstones from forming.

VITAMINS AND MINERALS - Such vit D, are considered essential nutrients—because acting in concert, they perform hundreds of roles in the body. They help shore up bones, heal wounds, and bolster your immune system. They also convert food into energy, and repair cellular damage.

ZOLOFT® - an antidepressant primarily used to treat major depressive disorders.

ZYRTEC® - used to treat cold and/or allergy symptoms such as sneezing, itching, watery-eyes, or runny nose.



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 in the pool, play sports, hang out with a friends, and even help out around the house and yard! Given many of the same opportunities as any other child, MTM children thrive and love life!

In light of that, in this section 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 explain that coverage is warranted for your child.



Nursing Care

Some insurance companies and many state assistance programs provide for in-home nursing. 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 (ST). 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 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. It is important to let your electric company know your child is electrically dependent.

Although the intent of this waiver program was implemented specifically for children like ours, some states have exclusion clauses, wait lists, or limited resources. It is important to speak with hospital staff/social worker immediately to apply for whatever Medicaid waiver program your child may be eligible for to ensure the best coverage possible. [How to apply for Social Security Disability Benefits with Myotubular Myopathy.](#)

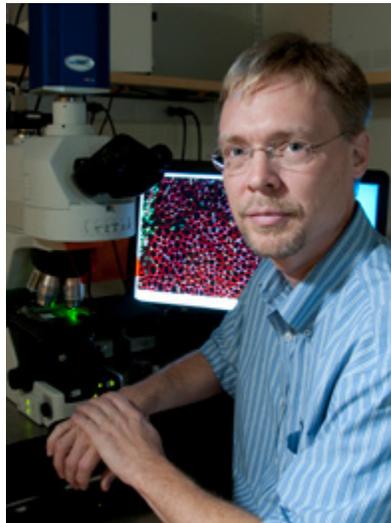




RESEARCH, TREATMENTS & REGISTRIES

Participation from our community is paramount to ensure the potential success of these therapies being studied. The research has advanced this far because of families like you were willing to share their family and medical information with these scientists. To view all research initiatives for our community, and to get involved please go to the [Get Involved](#) page on JFF's website. Also, another very important initiative is the [International Family Registry for Centronuclear and Myotubular Myopathies](#), the purpose of this registry is to create an investigator patient relationship in order to allow researchers to better understand CNM/MTM and locate subjects for clinical trials. If you or a family member are affected by CNM/MTM, [please register here](#) it will take only ten minutes of your time. There are two other registries also used by our community, [CMDIR](#), and [The Myotubular and Centronuclear Myopathy Patient Registry](#). You can find all three registries listed on the [Joshua Frase Foundation](#) website. Many of the subjects involved in the preclinical studies for our community up to this point, such as [RECENSUS](#), have utilized these very registries.

There are two biotechnology companies that are closing in on the first treatments for MTM. Audentes Therapeutics and Dynacure are working on two different potential treatments which have shown promise in the laboratory and pre-clinical data. Both companies have commenced clinical trials (human tests) and they are very interested in connecting with the CNM/MTM community and making a difference for our kids. Check out their websites at: www.audentestx.com, and www.dynacure.fr.





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 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. We offer two ways to honor...". There are far more than two ways, so saying "There are 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](https://www.joshuafrase.org) In Memorial page. We would also encourage you to register their information on the [International 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, date of birth and date they received their angel wings to info@joshuafrase.org.



Alison, Paul, Isabella and Joshua Frase



RESOURCES AND LINKS

Joshua Frase Foundation

To start with, you should begin by looking at the Joshua Frase Foundation website, www.joshuafrase.org. The JFF is the premier location for all things CNM/MTM. It has been funding research for over two decades and has funded more than 50% of the research dollars worldwide preclinical for gene therapy. Several of our families share their journey of perseverance and hope under [family stories](#). The website has tons of materials that you will find helpful. The Joshua Frase Foundation facebook page is, [Joshua Frase Foundation supporting Centronuclear and Myotubular Myopathy](#), it is a public page for updates on research initiatives and happenings within our community.



Facebook Support Groups

There is also a wonderfully active and supportive community on facebook, here are links to some of the most active closed groups; [CNM/MTM Families United](#) and [MTM/CNM \(Myotubular/Centronuclear Myopathy\) Family Support & Discussion](#). This forum is where parents can ask questions, discuss CNM/MTM topics and find encouragement from our global community.



Where There's a Will There's a Cure

The family of William Whiston formed Will Cure, www.will-cure.org to raise funds to support MTM medical research. Their efforts continue to 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, www.mtm-cnm.com held in the United States.

This is a grassroots event organized by a team of families providing a wonderful opportunity to connect with other families and leading researchers in person. The conference website and facebook group are great for both resources and family networking.

www.facebook.com/groups/mtmcmfamilyconference



Myotubular Trust

Anne Lennox and Wendy Hughes, two parents of children affected by myotubular myopathy set up the Myotubular Trust 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.



ZNM - Zusammen Stark E.V.

They are the association for myotubular and other centronuclear myopathies in Germany. Giving families a voice, sharing information about CNM and the latest research, and networking with other organizations. Founded on June 5th, 2015.

<https://www.facebook.com/znmstark/>



Physicians/Geneticists to Contact

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



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.

www.shrinershospitalsforchildren.org



Muscular Dystrophy Association (MDA) Clinics

The MDA, 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 utilize a multi-disciplinary 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. www.medicalert.org



Additional support groups for our global families.

Please visit the JoshuaFrase.org website to locate additional foundations, resources and support in your country.





GLOSSARY OF MEDICAL TERMS

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 healthcare team.

AIRWAY: The passage that allows air to move to the lungs.

ALVEOLI: Small air sacs in the lungs that give the tissue a honeycomb appearance and expands the surface area for the exchange of oxygen and carbon dioxide.

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: A condition where aveoli in the lungs have collapsed or do not open for air. Part, or all of the lung(s) that has collapsed, or developed atelectasis, do not participate in gas exchange and may be at risk of infection which can contribute to low oxygen levels. This can be caused by a blockage of the airways or by pressure on the airways from outside the lungs.

Bi-PAP: Also known as Bilevel Positive Airway Pressure, is a relatively small, quiet machine that provides non-invasive ventilation by creating air pressure and airflow that is in tune with your own breathing. 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 (O₂) and carbon dioxide (CO₂) in the blood, along with blood pH and bicarbonate level. It is a good indicator of adequate ventilation.

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 (O₂) is moved from air to body tissues to use by the cells and carbon dioxide (CO₂) 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 our 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: Inadequate or deficiency of cells causing underdevelopment or incomplete development of a tissue or organ. 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 the passive 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 or even normal 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.

Of note, individuals with MTM1 mutations are NOT at risk for malignant hyperthermia. Those with CNM caused by RYR1 mutation, however, are potential at risk for developing MH.

MANUAL RESUSCITATION “AMBU” BAG:

An Ambu® bag (a form of modified manual resuscitator) is a hand-held device that is used to manually provide breaths of air into the lungs, through a face mask or a connection to a tracheostomy, of patients who are not breathing adequately on their own.

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 breathe 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. Early engagement with a palliative care team is often extremely helpful for navigating the complex challenges faced in the care of individuals with MTM.

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).

PELIOSIS HEPATIS: an uncommon vascular condition characterised by multiple, randomly distributed, blood-filled cavities throughout the liver. The size of the cavities usually ranges between a few millimeters and 3 cm in diameter.

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: Anything that affects, occurs within or relates to the lungs.

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.

PULMONARY RESERVE: The maximum increase in minute ventilation that you can maintain without exhausting your respiratory muscles.

PULMONOLOGIST: A medical specialist who diagnoses and treats lung diseases.

RESPIRATORY FAILURE: A condition in which respiratory function is inadequate to meet the body's needs.

RESPIRATORY RATE: The number of breaths per minute that we take. An adult typically breathes 12-20 times per minute. Children breathe faster than adults (although the rate is age-dependent).

RESPIRATORY THERAPIST: A health professional who treats people who have breathing or cardiopulmonary problems.

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 (absence or partial seizure) or surge through the whole brain all at once (tonic-clonic seizure). A seizure can present differently, depending on where the seizure is coming from in the brain. Some people can have seizures where their whole body tenses, just an arm or leg twitch, to a seizure that just looks like they are staring. If you have concerns about potential seizure activity in your child, please speak with your healthcare provider.

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.

SUCTIONING: The process of removing secretions from the airway/tracheostomy by applying suction through a catheter.

TIDAL VOLUME: The amount of air we inhale with each breath.

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.

TRACH: Short for “tracheostomy”.

TRACHEA: Also known as “windpipe”, this cartilaginous and membranous tube descends from the larynx and branches into the left and right main bronchi.

TRACHEOSTOMY: A tracheostomy is a surgical opening in the windpipe (trachea), made by a surgical incision below the Adam’s apple (below the vocal cords). A tube is placed in the opening, and air goes in and out through the tube instead of through the nose and mouth. For some, a tracheostomy is short term. For others, it is long-lasting or permanent.

TRACHEOSTOMY STOMA: The opening in the neck where the tracheostomy tube is inserted. Also known as “trach stoma”.

TRACHEOSTOMY TUBE: A curved tube that fits into the tracheostomy stoma. It consists of an outer cannula and a flange that allows for tracheostomy ties to go around the neck to secure the tube in place. Some tracheostomy tubes also have an inner cannula and/or a cuff. There are many styles and sizes available from a variety of manufacturers.

TRACHEOTOMY: A surgical procedure that creates an opening for an artificial airway to maintain an individual’s ability to breathe.

VENTILATION: The exchange of air between the lungs and the environment, consisting of inhalation and exhalation.

VENTILATOR: A device for giving artificial respiration or aiding in ventilation.

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.

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.





DURABLE MEDICAL EQUIPMENT / DME

COUGH ASSIST: The Cough Assist provides noninvasive respiratory therapy that safely removes secretions from the airway passages. The Cough Assist provides positive pressure to the airway and then abruptly switches to negative pressure to loosen mucous. This positive to negative pressure simulates coughing to free secretions from the lungs and airway passages. Cough assist devices are used to reduce the incidence of respiratory infections. Phillips Respironics cough assist machine is lightweight, and relatively portable.



Phillips Respironics T70 Cough Assist



Phillips Respironics CA300 Cough Assist

CPT: Also known as Chest Percussive Treatment, can be done manual or mechanically. The mechanical method typically involves high-frequency chest wall oscillation which uses an inflatable vest that is attached to a machine. The machine mechanically performs chest physical therapy by vibrating at a high frequency. The vest vibrates the chest to loosen and thin mucus. This is often used in conjunction with cough assist, though not always. The Vest by Hill-Rom is one type of mechanical CPT. There are different vest sizes, and even simple wraps around the chest for our littler ones.

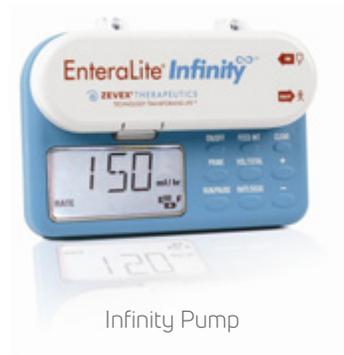


Hill-Rom Vest

FEEDING PUMPS: For those that are tube-fed, there are two main types of feeding pumps that our community has tended to use – the Kangaroo pump and the Infinity pump. These typically don't work as well for blended feeds (homemade food blends, not formula), however they are used often for formula based feeds. Both come with backpacks for both the feeding pump and its supplies.



Kangaroo Pump



Infinity Pump

HUMIDIFIERS: the Fisher & Paykel offer a lot of different models of humidifiers, as well as different types of chambers. There are two primary types of chambers. One that is manually filled to a fill line, typically by a care giver. The other is typically used with a sterile water bag (they typically come in 1L and 2L sizes), similar to what you would see in a hospital.



Fisher Paykel 850 System

NEBULIZERS help deliver breathing treatments. These can vary from simple saline to keep the airway moist to Albuterol or even heavier treatments to help nurse patients back to health. The Aeroneb line of nebulizers is something hospitals have started using, and have recently become available to some families via their DME company. Not all DME's carry them, but with a letter of medical necessity, sometimes you can get one. They are silent (ultrasonic), break the medicine into smaller particles which allow it to travel deeper into the lungs, and typically deliver treatments much faster than traditional nebulizers. Their biggest drawback is that their battery life has not been great. There are countless makes and models of the acorn style nebulizer. They can look plain, or they can be fun shapes for kids (like seals, etc). All of them typically come with the standard face mask, and there should also be a kit for use with a ventilator. Masks can also be purchased in fun animal shapes to help little ones feel more at ease with their treatments.

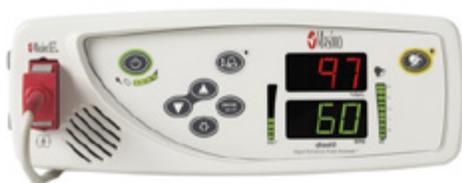


Aeroneb Pro Nebulizer



Kid-Friendly Nebulizer Kit

PULSE OXIMETERS monitor both the oxygen levels in the blood as well as the heart rate. They can be used full time (with the probe typically on the toe), or using a portable device that is used periodically on the finger tip. Masimo is a common brand of pulse-ox provided by DME companies, and uses a probe that is compatible with most hospitals. A portable finger tip pulse-ox can be used on the go, for periodic checks.



Masimo Rad-8 Pulse Oximeter



Portable Pulse Oximeter

SUCTION MACHINES: the DeVilbiss is a common suction machine used in our community, though not the only one available. The VacuAide model tends to be one of the quieter models out there, however, there are no silent models.



DeVilbiss QSU



DeVilbiss 7305

VENTILATORS: There are two primary ventilators used in the MTM community – Trilogy and LTV. Your pulmonologist can help guide your decisions as to which ventilator is best for your child. Trilogy by Philips Respironics is one of the two most used ventilators by MTM boys. It comes with an over the shoulder bag, and can be mounted on a wheelchair or stroller. The LTV ventilator is the other most used ventilator by MTM boys. It has a case that makes it able to be attached to wheelchairs. As an added bonus, currently the batteries in the Trilogy ventilators also work in the cough assist machines (both made by Phillips Respironics). With constantly evolving technology, new ventilators are emerging on the market that may not be shown here.



Trilogy 100*



LTV® 1150



TRANSPORTATION / CAR SEATS, STROLLERS + MORE



Cosco Dreamride Infant Car Bed

Infant Car Beds can be a good alternative for infants that are in the NICU and can't pass the car seat challenge to come home.



The Britax B-Agile / B-Safe Car Seat offers a larger recline than most car seats, and is part of a larger stroller setup. Offers a larger recline than most car seats.



Chicco NextFit, a convertible car seat with proven SuperCinch LATCH functionality and a LockSure vehicle belt-tightening system, easy-to-use adjustability. Also features a zip-off seat pad for machine washing and a breathable 3D AirMesh backrest.



Carrot 3 Booster Seat. Carrot makes a long line of special needs equipment. This is their booster seat.



Elite Tandem Stroller. The Tandem strollers offer additional space for equipment both in the secondary stroller seat and beneath.



Stokke Xplory. The Stokke stroller was a favorite among several parents. Baby is higher and faces parents.



Zippie Voyage. The Zippie Voyage can be purchased through a medical supply company and insurance may help cover it. It's designed for kids with special needs and sits on a Baby Jogger stroller base.



Special Tomato Jogger Stroller. Jogging stroller that is intended to work with the Special Tomato Soft-Touch Sitter chair.



Kid Kart Xpress

Seating can be placed forward or rear facing.



Snuggin Go. This positioning device helps younger kids with head and trunk support at early ages. Great for standard car seats and seating at home (bouncy chairs, etc).



Tumble Form Chair and Base. Tumble Form Feeder Seat with the Floor Sitter Wedge (typically sold separately). They also make a tray that fits well with this combination, allowing our children to play while in a seated position.



Special Tomato Soft-Touch® Sitter. Special Tomato chairs can be used similarly to the Tumble Form chair. Some brands works better than another.



GoTo Seat by Firefly/Leckey. Offers trunk support and modest head support. Can be used with the optional floor sitter accessory (allows for reclined seating). Can be used in every day activities where a typical chair is used.



EasyStand Bantam. A dual purpose stander and activity chair. The chair articulates from a seated into standing position and can be locked into any position in between. Great for kids who experience leg contractures which prevent full leg extension. It costs about as much as other standers and combines two bulky devices into one.



Leckey Squiggles Seating System. Leckey works for kids ages 1-5, as a mobile chair at home or in school. Provides postural support for younger special needs children.



Stander by Rifton. The Rifton stander helps strengthen core muscles and improve weight-bearing, weight-shifting and trunk control.



Rehatec Lasse Stander. Variable positioning through multi-functional adjustment of height, depth and tilt angle, provides safe supine positioning as well as interactive, supported vertical standing. For our friends in Germany.



DexBaby Crib Wedge. Crib wedges are good to have for elevating the head in their bed, or for general positioning during the day. Cloth cover also available.



Tumble Form Wedge. Wedges are often used in physical therapy, working on head control and play time.



Tumble Form Positioning Rolls. Often used for PT and strength development.



Mobility scooter by Tumble Form. Allows children to work on mobility and upper body strength.



Scoot Mobility Rider. Scooter that allows kids with a higher degree of trunk and head support have more freedom in movement



Summer Infant Deluxe Superseat 3-in-1 Booster, Activity, and Floor Seat. Another optional seating and play seat for those that have more head support.



Fisher Price Sit Me Up Floor Seat Frog. For those that have a little more head control, this can offer a play environment with a little support.



Hensinger Head Support. Soft foam wrap that goes around the neck and under the chin. Ideal for feeding, transport, sitting activities where additional head support is needed. Works with trachs/vents.



HeadPod Head Support System. The headpod offers freedom to turn the head while offering support. Attaches to standing frames and wheelchairs.



Abdominal Binder. An elastic abdominal wrap that can help support the torso in seating where harnesses may not be available.



Primo Eurobath. Bath chairs can take a while to get approved by insurance companies. This item has worked well for many parents.



Splashy Portable Bath Seat. Portable bath seat for toddlers and children. Has suction cup base to hold it in place and harness system to keep children from sliding.



Wenzelite Otter Bath Chair. Adjustable recline all the way to flat, elevated feet, straps to secure and wedges to hold head in position.



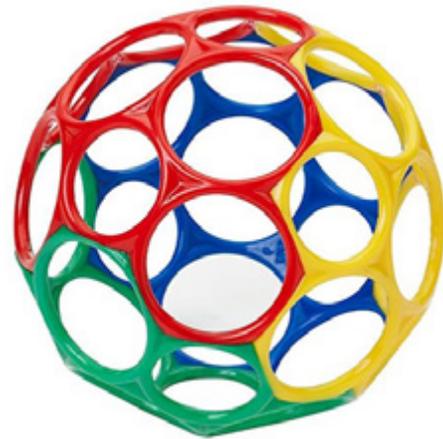
Sleep Safe Bed. A bed big enough for a teenager, with panels that can be removed for tubing and cords. Can be articulating.



HARD Pediatric Crib. Typically found in hospitals, can be found on Ebay for sale. Has hard-to-find drop rails, and head can elevate.



Babymoon Pod. Pillow that allows space for the ear if lying sideline, or elevates back of head if lying supine to help prevent flat spots.



OBall. The oball series gives lots of gripping opportunities to kids who may have a hard time grasping toys.



Z-Vibe, a vibrating oral motor sensory tool with interchangeable tip attachments, helps build oral tone and improve a variety of speech, feeding and sensory skills.



Baby Signing Time. Signing videos that can help teach kids (and parents) basic sign language.



Long Bendy Straws. Extra long bendy straws can be helpful for kids who can drink orally, but get fatigued easily lifting a cup.



Tobbi Dynavox, an eye tracking and eye control device that makes it possible for computers to know exactly where the user is looking. When eye tracking and eye control are combined with speech generating devices, communication opportunities are enabled for individuals with special needs.



Proloquo2Go is a communication device providing many natural sounding text-to-speech voices, 3 complete research-based vocabularies, over 10,000 up-to-date symbols, powerful automatic conjugations, multi-user support, ease of use and the ability to fully customize vocabularies.

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