

# The Care of Congenital Myopathy

## A Guide for Families



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# Foreword

Whether you've just received a diagnosis for your child or have known about it for quite some time, we trust that *The Care of Congenital Myopathy: A Guide for Families* will be a valuable resource for you. This guide represents the expertise and experience of medical professionals and families like yours from all around the world.

As you know, your child is unique. Not every scenario, treatment, or therapy presented here will apply to your situation. However, this guide will equip you to be a strong advocate while giving you much to think about and discuss with your medical team.

Throughout the following pages, you will find personal stories and photographs contributed by families who want to remind you that you're not alone on this journey. It is their hope that these words and images will provide you with helpful advice, encouragement, and a fresh perspective.



# Preface

This family guide summarizes the work performed by the International Standard of Care Committee for the Congenital Myopathies published in the *Journal of Child Neurology* (Ching H. Wang, et al. Consensus statement on Standard of Care for Congenital Myopathies, *Journal of Child Neurology*, 2012; 27 (3): 363-382). This task was supported by a grant from A Foundation Building Strength ([www.buildingstrength.org](http://www.buildingstrength.org)) and TREAT-NMD ([www.treat-nmd.eu](http://www.treat-nmd.eu)).

The Committee was composed of 59 experts from 10 medical disciplines including neurology, genetics, pulmonology, orthopedics, physical therapy and rehabilitation, gastroenterology, nutrition, and speech therapy. A two-step online survey was used to gather information on how doctors around the world care for people with congenital myopathy. A thorough review of published reports was completed to include the most up-to-date information. The group communicated by frequent email messages and periodic conference calls over the course of a year and a half, and formed this consensus recommendation after a 3-day workshop in May 2010 at Stanford University in California.

The purpose of publishing this care guideline is to establish a consistent approach to the diagnosis and clinical care of people affected with congenital myopathy and to identify areas that require additional clinical research to better future care. Visit <http://jcn.sagepub.com/content/27/3/363.refs.html> to download the Standard of Care document for free.

## HOW TO USE THIS GUIDE BOOKLET

This guide is intended for affected persons, families, and caregivers to understand the condition of congenital myopathy and optimize the care for people who are affected by this condition. Every effort has been made to use common language to make this document easy to read. In this booklet, the term “you” generally applies to the reader, who may be a caregiver or an affected person.

This guide provides an overview of congenital myopathy (CM) and how specific body systems are affected by CM. It details medical care needed for those with CM, including care from childhood and into adulthood.

The medical terms underlined in this guide are defined in the Glossary. Check the Table of Contents to find specific chapters within this guide as well as the Appendix, Resources for Families section, and Glossary of medical terms.

You may find it helpful to share this guide with health care providers, a spouse, your partner, school staff, other CM families, and other care providers. Your doctor may be able to help explain some of the points made in this guide if you have questions.

## DISCLAIMER

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

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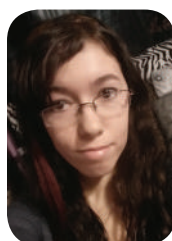
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# Introduction

You or someone you know has just received a diagnosis of congenital myopathy (CM). You may feel shock, denial, anger, or fear. You may be numb or overwhelmed by the news and the amount of information presented to you. You may be asking the question, "Why?"

Most people who are newly diagnosed with CM wonder what the future holds for them and their family. Your doctors and this Family Guide can help you see how the condition might affect your life. Due to the rarity of CM, your condition may be unfamiliar to the majority of medical care providers you will encounter, and the quality of care delivered often varies from one hospital or clinic to another. Therefore, understanding the disorder and its effects on the body will guide you in caring for yourself and help you advocate for better care.

The congenital myopathies are a group of rare muscle



disorders. The term congenital means "born with," but sometimes the symptoms are not immediately obvious at birth. New symptoms may also develop later in life. "Myo" means muscle. The suffix, "pathy" means a medical condition. Therefore, myopathy is a medical condition involving muscles. It typically results in muscle weakness and muscle fatigue.

Common symptoms and signs that occur in CM include:

1. weakness

2. low muscle tone or floppiness (hypotonia)
3. joint tightness (contractures)
4. breathing problems
5. feeding difficulties
6. bone abnormalities such as curvature of the spine (scoliosis)
7. delayed growth and motor achievements (milestones) such as rolling over, sitting, or walking

Knowing the signs and symptoms of CM can help you notice problems early on so that you can bring them to your doctor's attention. More often than not, these problems can be helped.

## What causes a congenital myopathy?

You may have felt worried that you somehow caused this congenital myopathy to occur, but know that you did not. Congenital myopathy cannot occur as a result of something you do, and you cannot "catch it" from someone else.

Congenital myopathy is caused by changes in genes that were passed through generations. Each cell within the human body contains many genes that work together to determine how the body is built. DNA is the material that makes up our genes. Changes in the DNA within the gene can lead to a difference in how the body grows and functions. CM is caused by a change in the DNA, called a genetic mutation or variant. These changes are what cause the muscles to be incorrectly built or not work properly, leading to the disorder's symptoms.

"We tend to assume that the medical staff will always know what is best. I think if we enter every situation realizing that each of us has useful knowledge and experience with CM, it helps to remind us that we (parents and those affected) can contribute in helpful ways and even educate others. We are all on the same team." - a mother of a child with CM

Genetic mutations can be passed down from parent to child (inherited), although they can show up spontaneously in a

family with no history of muscle disease. Geneticists can help find the exact nature or type of mutation you have. A geneticist or genetic counselor can review the chance of the condition happening again within your family. More information on genetics will be discussed later in this Family Guide.

### What are congenital myopathy subtypes?

There are many different types of CM. Doctors group CM into different categories called subtypes. Subtypes are named in two ways:

1. By microscopic appearance of the muscle cells, such as cores, nemaline rods, or central nuclei being seen
2. By genetic mutation

Doctors identify CM by observing the typical symptoms and by studying a piece of muscle tissue from the affected person. To view the muscle, a tiny piece of it is taken during surgery (muscle biopsy). The muscle sample is examined using a special process and microscope.

How is congenital myopathy different from dystrophy? In dystrophy, muscle cells are being broken down. In non-dystrophic myopathy, muscle cells are not contracting properly.

Another way to determine your subtype is through genetic testing, which can sometimes be done without a muscle biopsy when certain genes are suspected as being

the cause. Using saliva, blood, or a tissue sample from the affected person, a genetic testing laboratory can search for a genetic difference that is known or suspected to cause CM.

Sometimes a doctor will be able to make a diagnosis based on your symptoms or family history. However, most of the time, a muscle biopsy or genetic test is needed to confirm the diagnosis. A final confirmation by genetic testing is ideal.



The different subtypes, along with the gene mutation known to cause them, are listed in the Appendix.

### Why is there so much variation in congenital myopathy?



The way a muscle disorder affects someone varies from person to person. This is true even within a family with more than one affected member having the same genetic variant. The variation could be related to many things, like the age symptoms are first seen, the degree of muscle weakness, how the disorder affects other organs within the body, or how the condition is managed.

Some CM subtypes can be caused by changes in more than one gene. This means that some people with the same CM subtype may have a mutation in a different gene that leads to similar symptoms and the same muscle biopsy findings. Also, mutations in the same gene can cause different CM subtypes (see Appendix). Therefore, it is often helpful to have a geneticist, neurologist, or genetic counselor explain your form of CM and your genetic mutation to you, once it is known. Getting a correct diagnosis of CM may take some time.

### How can I get help managing congenital myopathy?

Ideally, a person with CM is best monitored by a team of doctors and specialists that are referred to as a multidisciplinary team. Depending on the CM subtype, symptoms, and complications, this team may include a:

- neuromuscular doctor (neurologist)
- genetic doctor (geneticist)
- lung doctor (pulmonologist)
- bone doctor (orthopedist)
- digestion specialist (gastroenterologist)



- nutrition specialist (nutritionist or dietician)
- oral/mouth doctor (orthodontist and prosthodontist)
- heart doctor (cardiologist)
- ear, nose, and throat doctor (ENT)
- occupational therapist
- physical (physio) therapist
- speech therapist
- psychologist
- rehabilitation specialist or physiatrist



Playing an instrument can be a source of accomplishment and joy.



Connecting with other families and people with CM can be a great source of information and support.

A good way to find the best team to care for you may be at a hospital with specialty care in muscle disorders. Explore all your options for the best doctors and realize they may be found at various medical centers.

It is important to remember:

- There are several different types of CM. Not all people with CM have all the symptoms or need all the care or treatments described in this guide
- There are similarities between people with CM; however, a person's course with CM is unique
- Care must be individualized



Family vacations can make for wonderful memories.



# 1

## Comprehensive Care

Medical care for individuals with congenital myopathy (CM) may require the involvement of multiple subspecialists. Sometimes those subspecialists work together in a multidisciplinary clinic, while in other cases, a family schedules separate appointments with each subspecialist.

It is important for the doctors taking care of you to help draft a care plan. This care plan may change over time and should prioritize the most pressing health concerns, their surveillance, and management. Most importantly, the care plan needs to be communicated and coordinated between the various subspecialists taking care of you. This section is divided into three topics: [diagnostic visits](#), [outpatient visits](#), and [hospital visits](#).

### [Diagnostic Visits](#)

The diagnostic visit is when your doctor (neurologist, neuromuscular specialist, or genetic counselor) explains the diagnosis in detail to you. You may feel overwhelmed with the amount of information presented to you. Your doctor should explain the muscle disorder, the probable outcomes ([prognosis](#)), and the problems you could encounter in words you can understand. You should also be given resources for support, such as a means to contact other individuals and families that have a similar muscle condition.

What you will learn at this visit will help you to predict problems ahead of time and to be prepared. You are encouraged to ask questions, take notes, and bring other family members or friends to this appointment. Commonly, a person will need some time to process the new information before thinking of questions to ask. If one visit is not enough for you,

schedule another visit with that provider, or another specialist that may be able to explain things to you in ways you can understand. The doctor or specialist should allow enough time for discussion during this visit. From this point forward, you will likely have regularly scheduled appointments with various specialists.

### [Six Key Topics for Discussion During the Diagnostic Visit](#)

1. **Diagnosis**—Your doctor will explain the specific subtype and gene mutation (if known) causing your CM. Your doctor should also explain the consequences of muscle weakness, such as difficulties with movement, breathing, and feeding. Your doctor should also describe how the disorder affects other body systems.
2. **Prognosis**—There is a wide range in how people are affected by their CM, so there is a wide range of outcomes. In general, the first few years of life tend to be the most serious because of breathing difficulties, feeding problems, and regular illnesses. Illnesses tend to be severe and will worsen breathing and feeding difficulties. Life expectancy of CM is very difficult to predict, even within a family. Predicting life span is a topic to discuss with your medical team. Life span and quality of life has improved because of new technologies, especially related to the management of breathing issues.
3. **Genetics and risk of having another child with CM**— If the genetic cause of your CM is known, a genetic counselor can explain what the chances are of other family members having the same CM. Even without knowing the genetic cause, you can be given an estimate of the risk it could happen again. CM

can run in families, though it could also happen spontaneously. It is possible that other family members could have a milder form of the disorder or could develop CM later in life. If you have concerns about this, please discuss it with a geneticist or genetic counselor.

4. Care plan—The main goal of a care plan (also called a treatment plan) is maintaining function. To accomplish this, you will learn to:

- a) Anticipate problems before they happen, to head them off
- b) Promote maximum movement and growth of your body

It will be hard to remember everything, and that's where a care plan comes in. A care plan can help you to understand how your days will be patterned to meet the needs associated with CM. A daily care plan can help to organize the cares needed every day, such as respiratory treatments, special dietary needs, or medications. Pulmonary and nutritional concerns are a particular priority, especially during the early years of life.



A monthly or yearly care plan can help you coordinate care over a longer time period and would include follow-up clinic visits. It is ideal to have both a “well” care plan and a “sick” care plan. Your plan will likely need to be adjusted as your needs change.

Following a medical appointment, ask for a report called an “After Visit Summary.” This visit summary should include information like height and weight, vital capacity, or degree of spinal curvature (if you are monitored for scoliosis). You can use the information from your After Visit Summary to judge how well your care plan is meeting your needs and if changes are warranted.

5. Support groups and other resources—It will be helpful to receive information about family support groups (online and in-person) and educational resources, like this guide. Meeting and sharing your experiences with other families can be comforting and makes you realize you are not alone. For a list of family resources, see the Appendix.

6. Research opportunities—There are opportunities to participate in research studies to expand understanding of your subtype and advance treatment discovery. You can find a list of some places to learn more about research studies for congenital muscle disease in the Appendix.

### Outpatient Clinic Visits

You will be scheduled for follow-ups with specialists at regular intervals, likely varying from 3-6 months during the early years of your CM or when symptoms may require closer monitoring. During each visit, a nurse will measure blood pressure, heart rate, breathing rate, weight, height, and head size (during infancy). A measure of the length of the bone in the forearm (ulnar) can be used instead of height in some situations, such as when a patient has a curved spine or is unable to stand.

### What Should be Evaluated During These Visits?

Lungs— Preventing lung infections is crucial. Lung infections, called pneumonia, can hinder growth and lead to hospitalization. In some cases, these infections can be deadly. How well your lungs work (lung function) will be routinely monitored through breathing tests called pulmonary function tests (PFT). In addition to getting the typically recommended

vaccines, obtaining flu and pneumonia vaccines are strongly encouraged.

**Nutrition and growth**—Children with CM will not follow the typical growth patterns. If your child fails to gain weight, loses weight, or gains too much weight, then it should be brought to the attention of your doctor. Warning signs of an eating problem include frequent choking or coughing when eating or drinking, vomiting/reflux, and constipation. These issues should be brought to the attention of your doctor right away.

**Social**—A social worker can help you with the more practical aspects of living with CM, such as health insurance, disability services, social services, equipment and supplies, educational needs, and finding emotional support.

**Growth/Development**—Many with CM will have difficulty achieving motor milestones. These needs can be addressed by physical, occupational, and speech therapists. Most children with CM do not have problems with their intellect, such as understanding language and problem solving.

**Skeletal system**—You may develop an abnormal curvature of the spine called scoliosis or tightening across the joints, called contractures. These malformations will need to be monitored

Routine liver function tests and abdominal imaging studies are recommended for all XLMTM patients

by a rehabilitation specialist and/or orthopedic surgeon. Severe scoliosis often requires surgery to prevent the abnormal curvature of the spine from causing breathing problems. Also, people with CM are at risk of having thin bones, so, regular calcium and vitamin D supplementation is encouraged.



**Emotions/Behavior**—Mood or behavior troubles may occur in children that realize the differences in themselves compared to other children of similar age. Addressing these issues with the help of a child psychiatrist or psychologist can be beneficial. Also, as a person with CM goes through the various life stages, they may need more emotional support. Friends or family are not always available as supports or may not be enough. Treating mental health issues is just as important as treating physical health issues, and should not be ignored.

Caregiving is stressful emotionally and physically. Caregivers are also at risk of mental health concerns and should not ignore their own needs. Sometimes, talking through these issues is enough, but when talking is not effective there are medications that can help you to feel more like yourself.

**Ears, nose, and throat**—Many people with CM need monitoring of their ears and hearing ability because

fluid can tend to remain trapped, causing discomfort, mild hearing loss, and possible ear infections. Some people with CM have enlarged tonsils and/or adenoids that can cause airway obstruction, so it is recommended to see an Ear, Nose, and Throat (ENT) specialist for an evaluation.

**Heart**—Most CMs are not associated with abnormal electrical heart rhythms (arrhythmias) or abnormal heart contraction (cardiomyopathy, also known as heart failure). Sometimes heart problems may be associated with some forms of CM, this is rare. It is more likely that heart problems occur because of severe lung problems. The cardiologist will likely perform at least one heart evaluation, consisting of an electrocardiogram (ECG or EKG) to measure the rhythm of the heart, and a heart ultrasound (echocardiogram).

### Hospitalization

You may need to be hospitalized suddenly for various reasons.

You should make sure your primary doctor and your regular specialists are aware of your hospitalization. Reasons for hospitalization include:

1. Severe breathing problems—During acute illness, such as having an infection, you may need to be placed on a breathing machine (ventilator).
2. Heart problems—You may need medications to help the heart beat harder or stop an abnormal heart rhythm.
3. Feeding problems—“Failure to thrive” means there is poor weight gain or too much weight loss in children. This is also referred to as being undernourished. Your doctor may suggest feeding through a tube that enters at the nose (called a nasogastric or NG tube) and goes into the stomach as a temporary solution. If your child does not gain the ability to swallow safely or if there is concern that he or she is not eating enough calories each day, then a stomach tube (called a gastrostomy tube, or G-Tube) may be inserted. These two most common feeding alternatives also work well for adults with CM that need assistance for proper nourishment.
4. Dehydration—Becoming dehydrated is a serious condition and can happen within just hours, especially in someone of low body weight. During bouts of diarrhea, vomiting, common colds, fevers, physical exertion, or sweating, body fluids are lost at a rapid rate and serious dehydration can occur. Dehydration can come on quickly (acute) or have a gradual onset and continual presence (chronic). Gradual or chronic, low to moderate dehydration often presents with:

- headaches
- tiredness
- sunken appearing eyeballs
- constipation
- dry skin
- minimal, dark colored urine
- sticky saliva
- irritability
- lack of concentration
- low blood pressure



For some, there is diminished or no self-awareness of being thirsty or dehydrated. Rehydration drinks, like Pedialyte or Gatorade, help better than plain water because they also contain sugars and salts (electrolytes) in the right measure to

replenish what the body is missing.

For sudden, severe dehydration, watch for the symptoms mentioned above and also for signs of:

- unusual sluggishness (lethargy) and extreme tiredness
- pounding or irregular heart beat
- sudden changes in breathing
- dizziness
- delirium
- fainting and loss of consciousness

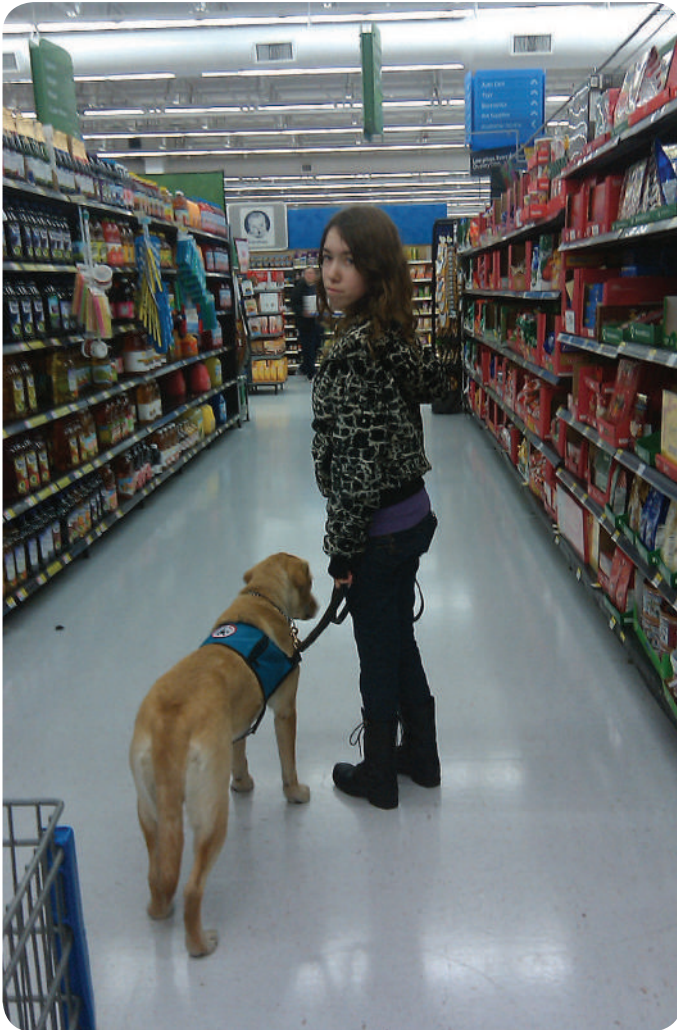
This type of sudden, severe dehydration can be life-threatening and often requires immediate replenishment of intravenous (IV) fluids through a vein at the hospital. A trip to the emergency room will be required if you are unable to rehydrate the person within a couple of hours on your own.

Ideally, homemade rehydration drinks should contain:

- starches and/or sugars as a source of glucose and energy
- sodium/salt
- potassium

5. Malignant Hyperthermia (MH)—This is a rare condition in which some people with CM who are undergoing surgery and given certain types of anesthesia or muscle relaxants can develop very high fevers and a high metabolism. It is a dangerous condition and requires close monitoring. If you need to undergo surgery, your neuromuscular specialist should discuss with you the risks and benefits of the procedure, including the risk of malignant hyperthermia when given anesthesia, Succinylcholine and halothane are the types of anesthesia that should be avoided. The anesthesiologist should use a “clean technique,” which means that the equipment was cleaned of all traces of anesthesia used on a previous patient. They should also have the MH rescue medication, dantrolene, on hand in the surgery room. Make sure your surgery team knows you may be at risk for MH.

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All CM subtypes should be considered a possible risk for MH. Mutations in the ryanodine receptor (*RYR1*) gene are known to be associated with malignant hyperthermia.  
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# 2

## Neuromuscular Concerns

Congenital myopathy is a neuromuscular disorder affecting muscles under voluntary and involuntary control. While weakness is the primary symptom, congenital myopathy can affect many different body systems and lead to additional health concerns and symptoms.

### Weakness, Pain and Fatigue

People with CM experience muscle weakness, pain, and fatigue. The exact cause of the muscle pain is unknown, but may be caused by the muscle disease itself, stiffness in the joints, thinner bones, or joint deformities that develop over time. Muscle soreness is commonly the result of physical exertion, such as prolonged standing or writing. Fatigue will occur more easily and sooner for a person with CM due to the underlying muscle weakness.

Generalized or chronic muscle pain and soreness can be treated by:

- Regular stretching
- Manual mobilization of affected areas
- Massage
- Over the counter medications
- Prescription medications which block neuropathic pain, such as Gabapentin

Fatigue is very common in people with CM. This could be due to burning more calories than normal because of the differences in their muscle function. Supportive devices like walkers, scooters, and wheelchairs may prevent fatigue and promote greater mobility and independence in daily activities. Evaluation and physical therapy is recommended not only for

treatment of muscle pain, fatigue, and reduced endurance but also to provide guidance when considering mobility equipment. Please go to the Appendix for suggestions to help you find mobility equipment on the Resources page.

### Weakness of Breathing Muscles

Some people with CM are born with weak breathing muscles and need the assistance of a breathing machine (mechanical ventilator)

More information on breathing concerns can be found in the Respiratory Management chapter of this guide.

either temporarily or permanently starting at birth. Others with CM develop breathing muscle weakness over time. This is a gradual process with breathing issues starting first at night in many cases.

### Weakness of Face and Swallowing Muscles



Some people with CM experience weakness of the face and swallowing muscles. Weakness in these muscles can cause sucking, eating, drinking, and speaking difficulties.

If a swallowing problem is identified, there are some options to consider. These options will be explained

in more detail in the Feeding, Nutrition, and Oral Care chapter found later in this guide.

It is suggested that caregivers become trained in CPR and other rescue techniques, like those for choking.

### Weakness Leading to Dysfunction of Intestines

The small intestine, large intestine (colon), rectum, and anus can also be affected by the muscle weakness. You could experience persistent or frequent occurrences of constipation, cramping, and/or diarrhea due to a lower level of physical movement and activity. For detailed information about gastrointestinal concerns, please see the chapter on Feeding, Nutrition, and Oral Care found later in this guide.

### Weakness or Dysfunction of Ear Muscles

Some people with CM may have eustachian tube problems associated with muscle dysfunction. The eustachian tube is a narrow channel that connects the middle ear with the nasopharynx (the upper throat area just above the palate, in back of the nose). The eustachian tube is approximately 1½ inches long in adults, but much shorter in children. There are four muscles controlling the function of the eustachian tube. When these muscles are not working properly, symptoms can occur such as:

- Fluid retention in the middle ear leading to muffled hearing and higher risk of ear infections
- Painful pressure in the middle ear because there is less ability to equalize pressure

Since children may not complain of these symptoms or be able to describe what they are experiencing, you should have regular examinations of your child's hearing and ear pressure. For persistent cases, surgical insertion of tiny tubes in the affected person's eardrum usually solves the problems associated with eustachian tube malfunction.



### Weakness and Incoordination of Muscles Used in Speaking

Weakness of the diaphragm, throat, mouth, soft palate, and facial muscles may also lead to difficulty with speaking. More information on speech concerns can be found in the Feeding, Nutrition, and Oral Care chapter of this guide.

### Weakness Leading to Orthopedic Complications

Orthopedic problems, such as dislocation of the hips, joint tightness, or an abnormal curve of the spine are often encountered as a result of muscle weakness. More information on orthopedic concerns can be found in the Orthopedics and Rehabilitation chapter of this guide.

### Weakness and Deep Tendon Reflexes

During medical visits, your doctor has probably checked your reflexes using the small rubber-tipped reflex hammer. In CM, a person's deep tendon reflexes can be low (hyporeflexia) or absent (areflexia). Joint hypermobility, another common occurrence in CM, means having greater than normal flexibility.

### Weakness in Eye Movement or Eyelids

Weakness of the eye or eyelid muscles can occur in people with certain types of CM, like Centronuclear Myopathy. Fortunately, most children do not develop a vision problem (like double vision) because the brain is able to adapt over time.

Weakness of the eyelid muscle can cause the lid to droop or appear partially closed (ptosis). Sometimes it will hang down low enough to cover the pupil and this can block vision in that eye. Surgical correction of a droopy eyelid can be an option to improve vision.

In some people, the eyelid may not close all the way during sleep. When this happens, there is a risk of scratching the clear



outer layer of the eye known as the cornea. A scratched cornea is painful and can impair your vision. Eye lubrication with a gel while sleeping can help prevent scratching and dryness of the eye.

Weakness of the eye muscles can make it difficult to look in various directions without moving the head (ophthalmoplegia). If some eye muscles are weaker than others, the eye may not appear to look straight ahead when the person is looking forward. Unfortunately, this condition cannot be corrected.

In rare cases, some people with x-linked myotubular myopathy and female carriers of the disease have developed cataracts. It is something that patients need to be aware of and screened for, as a precaution.

### Weakness of Heart Muscle

Heart problems are not common in CM; however they can occur in some cases either due to a primary heart problem or due to underlying breathing issues. If the muscle disorder affects the heart (which is a smooth muscle), it is known as cardiomyopathy. In cardiomyopathy, the heart muscle becomes enlarged, thick, or rigid. In rare cases, the muscle tissue in the heart is replaced with scar tissue.

As cardiomyopathy worsens, the heart becomes weaker. It's less able to pump blood through the body. When the heart cannot beat with enough power to supply the body with blood it is called heart failure. In turn, heart failure can cause fluid to build up in the lungs, ankles, feet, legs, or abdomen. Heart failure has been found only in rare cases with mutations in the following genes:

- *ACTA1* (*actin α1*)
- *DNM2* (*dynamitin 2*)
- *TPM2* (*tropomyosin 2*)
- *SEPN1* (*selenoprotein 1*)
- *TTN* (*Titin*)

The heart muscle could also have an abnormal electrical rhythm, causing it to beat irregularly. This is called an arrhythmia. Brief heart failure and irregularities in the electrical activity of the heart have both been reported in some people with nemaline myopathy.

CM patients without symptoms should be screened by a cardiologist every two years to make sure the heart is working properly. More frequent evaluations may be recommended if you have obvious symptoms or known heart abnormalities.

### Progression of Weakness

The degree of weakness can change over time, either improving or worsening. In general, after the first few years of life, gradual improvement in muscle strength can be seen, and sometimes maintained into adulthood. An improvement in strength is more likely to occur in those who participate in physical therapy and other physical activities to the greatest extent possible.

In mid to late adulthood, muscle weakness may gradually worsen, as it does with everyone as they age. With age, breathing concerns may show up and scoliosis may develop or worsen. If walking is achieved, that skill is usually not lost until much later in life, although walking may become more tiring or difficult with advancing age.

Some people will experience worsening of symptoms in the teen years or early adulthood. Walking can also sometimes be affected by a rapid growth spurt, such as during puberty, when the muscles may take longer to catch up with a growing skeletal frame.

Life span varies based on the CM subtype, genetic mutation, severity, and ultimately, on breathing function. Unless there is severe respiratory failure during infancy, most children with CM live into adulthood.

### Brain Involvement

Children with CM usually have normal intellect. In one report, three children with *ACTA1* mutations were found to have a developmental delay in word comprehension. Another report found two children with *BIN1* mutations to have mild mental retardation. In addition to skeletal muscle weakness. More studies are needed to determine if any relationship exists between cognitive delays and CM.



# 3

## Respiratory Management

Good airway management and treatment may enable an infant, child, or adult to have a better quality of life and a longer life. Breathing problems are sometimes more predictable with some types of CM, but of course, everyone is unique.

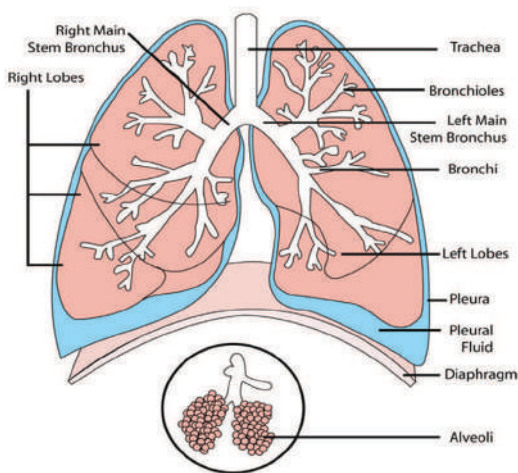


Image credit - <http://www.interactive-biology.com/3331/how-do-we-breathe-lungs-and-pleura/>

The diaphragm muscle, muscles between the ribs (intercostal), and abdominal muscles are all used for breathing. The breathing muscles allow you to inhale oxygen (O<sub>2</sub>) and exhale carbon dioxide (CO<sub>2</sub>). These muscles are often weakened in CM.

With weakened muscles, a person is not able to take in as much air (breath volume). There are some problems that result from this. First, a child's lungs may not grow and develop properly when there is not enough air going into all parts of the lungs. Second, decreasing breath volumes can cause a build-up of carbon dioxide. If carbon dioxide builds up in the body, it can lead to changes in blood pH (acidosis). This build-up

of carbon dioxide is more likely to happen initially at night because we naturally breathe less deeply when sleeping. Third, coughing ability is reduced. Coughing is important to clear the airway and prevent pneumonia, as well as recover from lower respiratory infections.

Breathing issues can be treated by providing a person with breathing support devices, which can take over the effort of breathing to increase breath volumes or assist with coughing. In a person with CM, the ability to breathe can be impaired immediately after birth or can develop later in life. Muscles used for breathing in some people with CM may be weaker than the muscles of the arms and legs, which can often disguise the breathing problem. Therefore, breathing function should be monitored early and often by a pulmonary doctor or other specialist with expertise in managing breathing muscle weakness, such as a respiratory therapist, neurologist, rehabilitation medicine physician, or physical/physio therapist.

### Weakness of Breathing Muscles and CM Subtypes

The weakness of breathing muscles is the most important factor to predict life span.

Severe respiratory impairment requiring breathing support from birth is common in some forms of CM. In some infants with CM, the first year of life is when they are most vulnerable to breathing failure. They therefore require careful breathing management which may help to improve breathing function and prevent breathing problems.

However in others with CM, breathing problems may occur over time, or during other physical changes such as during the

development of scoliosis, during or after the loss of mobility, or during periods of rapid growth such as in puberty. Weakness of the intercostal muscles between the ribs can lead to loss of movement and stiffness of the rib cage, eventually misshaping the chest. The chest may appear sunken (pectus excavatum) or narrow. This can impair lung capacity and should be treated early with breathing therapy, which will be explained later in this section.

Someone with CM will benefit from being seen by a team of doctors that specialize in treating children or adults with neuromuscular conditions like CM. Most importantly, caregivers need to be well trained in techniques to manage the airways in someone who develops breathing problems associated with their CM.

### What is Ventilation and Why is this Important to Understand?

Ventilation is the process whereby your chest wall expands during inhalation, air enters the lungs, oxygen goes into the blood, and carbon dioxide is exhaled. Proper ventilation is often difficult for people with CM because of weakness of the muscles involved in breathing. Scoliosis can impair chest expansion and then lead to problems with ventilation, so breathing management will require careful follow-up between an orthopedic surgeon and a breathing specialist.

Decreased lung volumes lead to carbon dioxide build-up over time. When there are ventilation problems, you may see the carbon dioxide level go up before you see the oxygen level get dangerously low. This is why measuring the carbon dioxide level in people with CM is just as important as measuring oxygen levels.

### What are the Symptoms of Breathing Difficulties?

Your breathing specialist will monitor your breathing abilities regularly. Recognizing the early symptoms of respiratory difficulty is very important. If you are concerned about breathing, please call your specialist. If the issue is urgent, go to the emergency room.

These are common symptoms that may indicate respiratory problems:

- Fatigue
- Appearing anxious
- Weight loss or poor weight gain (sometimes called “failure to thrive” in infants)
- Frequent lung infections
- Weak cry in the very young
- Shortness of breath
- Irregular, forced, or rapid breathing (rib flare or using the tummy more when breathing)
- Weak cough
- Frequent choking on saliva
- Coughing and choking during feeding
- Poor appetite
- Poor growth

Symptoms or warning signs of high carbon dioxide during sleep:

- Periods of no breathing during sleep greater than 15-20 seconds (apnea)
- Frequently waking up either partially or fully at night, or restlessness during sleep
- Difficulty waking in the morning
- Feeling groggy or sleepy during the day even though you had enough hours of sleep
- Difficulty concentrating or poor school/work performance

### Helpful Equipment to Have at Home for Most Subtypes

- Suction machine
- Carbon dioxide and oxygen monitors
- Nebulizer
- Cough assistance
- Ambu® Bag Resuscitator
- Percussive device (cupping device or vest)
- Humidifier
- Vaporizer

Also, obtain back-up machines to cover breakdowns and a generator for when the power goes out.

Tip: notify your power company that you have life-sustaining equipment at home so that your home is a priority residence for getting or keeping power on.

- Headaches or irritability
- Poor appetite in the morning or throughout the day
- Nausea
- Loud snoring (older CM patients)
- Sleeping problems or nightmares
- Bluish tinged tongue or lips

One or more of these symptoms could indicate carbon dioxide build-up, and should be discussed with your doctor or respiratory nurse.

### How Can We Monitor Lung Function and Breathing Muscle Strength?

Pulmonary Function Tests (PFTs): The most common PFT is called spirometry. It is a simple, non-invasive test that measures the amount of air that can be exhaled after a maximal inhalation (also called forced vital capacity, or FVC).

The FVC is often converted into a percent of predicted (FVC % predicted). This value reflects the percentage of air volume exhaled compared to an unaffected person of similar height (or length), sex, race, and age.

The forced vital capacity is usually measured while sitting. A difference in FVC % predicted when lying down compared to sitting may indicate increased diaphragm muscle weakness. Based on this result, your doctor may recommend a sleep study to check for decreased breathing at night even if the sitting FVC % predicted is within the normal range.

The PFT can be done in children six years of age or older. However, beginning the test in children as young as age four may help teach them how to perform the test.

Traditional spirometry and other tests that rely on patient exertion may be ineffective or inconclusive for individuals with severe respiratory weakness.

You may also be asked to cough into a tube to measure the strength of your cough. This is called peak cough flow. Maximal inspiratory pressure (MIP) and maximal expiratory pressure (MEP) are also important breathing muscle strength measures. These tests are done every six months or once a year, usually by a respiratory therapist working with a pulmonologist.

Oxygen measurement: Pulse-oximetry, also called pulse-ox, uses a painless, small sensor that is attached to a finger or toe to detect the amount of oxygen in the blood. This is an easy way to find out if the body is getting enough oxygen. If the measured oxygen level is less than 95%, this indicates a problem such as secretions in the airway or lowered breath volumes. Do not offer oxygen to someone with CM with oxygen levels below 95% unless you have already considered the following:

- Use a cough assistance machine or manual cough assistance (see Appendix for instructions) to help remove secretions or mucus plugs. Follow up with suctioning to remove loosened mucus, as needed.



### Inappropriate Oxygen Use

After being admitted to the ER of a reputable children's hospital for respiratory distress, my daughter was getting over 10 liters of oxygen with no BPAP support. Her oxygen levels were good (97% or so), but obviously no one was thinking about carbon dioxide. My daughter

was very young and we were still new to the diagnosis and her care, so we had no idea this treatment was all wrong for her.

Once we saw an actual doctor (about 30 minutes after getting admitted) he recognized the problem of using oxygen and not monitoring her carbon dioxide levels. He got her on BPAP right away and ordered the oxygen to be turned way down, but that was not before her carbon dioxide levels had hit the 90s and she was acting "drunk." That was double the acceptable carbon dioxide level.

It took about an hour or more on BPAP for her to get back to appropriate carbon dioxide levels. This was before we had a BPAP machine at home and when we didn't know enough about her respiratory care, but now I know much better. It stinks that this happened to us and I'll do anything to prevent it from happening to others.

- If possible, measure carbon dioxide level to determine if the individual requires ventilator support or a change in ventilator settings. In some regions, capnographs are not readily available for home use, but if your doctor ordered oxygen, you should have a way to measure both oxygen and carbon dioxide levels at home. (See capnography explained below.)
- Rule out a lung infection (pneumonia), if appropriate.

Carbon dioxide measurement: Capnography is a measure of the amount of carbon dioxide in your blood. A capnograph is a device that painlessly measures the amount of carbon dioxide in your breath, which is closely related to the level of carbon dioxide in the blood. It can be measured using a mouth piece or a nasal cannula that fits into the nostrils.

In a person with a tracheostomy and ventilator, carbon dioxide can also be measured by adding a capnostat, which is a plastic piece that fits into ventilator circuits.

A capnograph is sometimes prescribed by a breathing specialist to measure carbon dioxide levels at night either in the hospital or at home.



The CM standards of care include having a sleep study regularly, with O<sub>2</sub> and CO<sub>2</sub> levels measured. Sleep studies, like the one pictured above, should begin as soon as CM is diagnosed, even before a person has symptoms of disordered breathing.

Another way to measure carbon dioxide in your blood is to wrap a small sensor around a finger to measure carbon dioxide levels just under the skin (transcutaneous measurement).

As discussed earlier in this section, elevated levels of carbon dioxide are dangerous and should be treated with ventilator support and/or airway clearance.

Do not just give oxygen to make the oxygen saturation go up without addressing the root cause of the decrease first. In a breathing emergency, offering oxygen may be beneficial as long as there are not secretions in the airway.

Invasive oxygen and carbon dioxide measurement: Arterial blood gas is an analysis of blood oxygen and carbon dioxide levels obtained from your blood. It is usually performed in a hospital, clinic, or sleep lab setting.

Sleep study: Because most breathing problems first manifest during sleep, a sleep study might be required, especially if you have symptoms of elevated carbon dioxide or have an abnormal FVC % predicted.

Doctors call this type of test polysomnography or PSG, but it is more commonly known as a sleep study. It is an overnight procedure in which you sleep in a sleep lab or hospital. A technician monitors many factors, including oxygen level, carbon dioxide level, pulse, brain waves, heart rhythm, and chest movements. This is the best way to detect poor ventilation or sleep-disordered breathing like sleep apnea, in which people stop breathing for short periods of time during sleep. Obstructive sleep apnea in CM is usually caused by weakness of the throat muscles. Those with CM are advised to have routine sleep studies to catch problems early.

Sleep studies can help determine if you need to use bilevel ventilatory support. This type of breathing support includes a breathing machine that acts like a pair of bellows and will take over the effort of your breathing at night. A sleep study can also help determine if your ventilation choice is effectively supporting your breathing needs at night or if the settings need to be changed on your breathing machine.

Equally, if someone is over-ventilated because the settings on their breathing machine have been set too high, it can

reduce their CO<sub>2</sub> levels by too much, which may cause similar symptoms to sleep disordered breathing. It is therefore important that you have regular sleep studies, even if you already have overnight ventilation.

## Ways to Assist Breathing

There are two basic modes of assisting with breathing:

1. Non-invasive
2. Invasive

### Non-Invasive Ventilation

Breathing support that is non-invasive provides air flow from a ventilator or breathing machine to the lungs using a:

- Mouth piece
- Nasal mask
- Nasal pillows
- Face mask
- Helmet mask

A ventilator is a simple device that acts like a pair of bellows to help you improve your breathing. The ventilator delivers a gentle pressure which helps your lungs to expand.

There are a variety of methods that deliver either a certain pressure of air, or a specific volume of air. Some examples include:

- Bi-level positive airway pressure [BPAP]
- Continuous positive airway pressure [CPAP] – not appropriate for hypoventilation in CM
- Intermittent positive pressure ventilation [IPPV]
- Proportional-assist ventilation [PAV] which provides flow and volume assistance with each breath
- Ventilator which delivers a breath with pressure or a set volume.

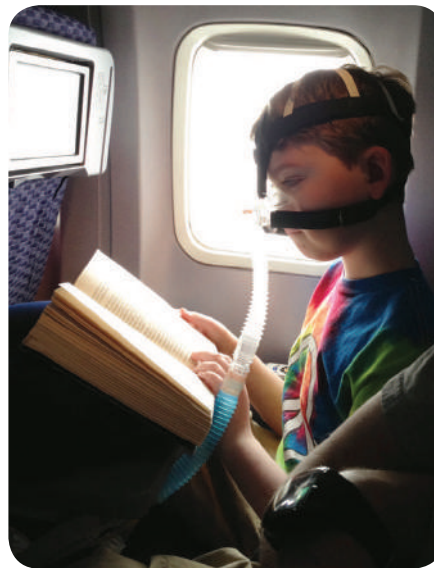
These devices are explained below:

**Bi-level positive airway pressure (BPAP):** BPAP ventilators deliver air through a tube that connects onto a mask covering the nose only, or covering both the nose and mouth. It assists by pushing air into and allowing air out of the lungs. The mask can easily be removed when the breathing machine is not in use.

Non-invasive means it does not penetrate the body. Invasive means requiring a surgical procedure or device that penetrates the body.

BPAP helps correct low oxygen levels and high carbon dioxide levels, and gives breathing muscles some rest. BPAP can be used during night-time sleep, naps, when sick, off and on throughout the day, or all day, depending on your needs.

The BPAP is the most common treatment for poor ventilation in CM. It can help keep the lungs in good health and the rib cage flexible. It can also help to improve quality of life such as increased energy levels, better concentration, and can often prevent breathing problems such as chest infections.



Note: BiPAP® is the name of a portable ventilator manufactured by Respironics Corporation; it is just one of several ventilators that can deliver BPAP. Your respiratory care specialist will determine the appropriate settings for the machine in order to properly ventilate the user.

Continuous positive airway pressure (CPAP): CPAP is a treatment that uses continuous air pressure to keep the back of the throat open. It is not appropriate for people with neuromuscular conditions because it does not provide enough

assistance for breathing in and it actually makes breathing out even harder for them. It is designed for people with sleep apnea only. It will not treat hypoventilation due to weak respiratory muscles.

**Sipper vent:** This device is positioned near the face with an angled mouthpiece connected to a ventilator. The user closes their mouth around the mouthpiece when they feel the need for an assisted breath. It provides a volume of air for talking and breathing at the same time.

**Mechanical ventilator:** This option can be used with a nose, or

nose/mouth mask, or through a tracheostomy tube.

Side effects of using non-invasive ventilator devices include skin irritation, bloating, reflux, and dryness. Many of these problems can be resolved. Some suggestions are below for managing these side effects and you may want to ask your specialist or others with experience using non-invasive ventilation for further advice.

Mask leaks and eye irritation can be alleviated by changing your mask to a better fit, putting dermal gel patches to “plug” any gaps in your mask, putting eye gel or drops in your eyes, or



wearing an eye mask.

Skin irritation and sores can sometimes occur due to your mask being too tight, a poor fit, or an imperfect design for your face. This may be resolved by providing you with a different mask which is better fitting, or by providing you with two types of masks for alternating use; applying creams or dermal gel pads to prevent pressure sores or dry skin, or loosening the straps on your mask slightly. There are many types of

interfaces to choose from, so you should be able to find a mask that is comfortable. Be patient, as it may take months to find and get used to a new interface.

Bloating caused by air getting into the stomach may be prevented by adjusting the pressure settings on the machine to reduce air trapping, or by changing to a different mask. Some medications may be able to help prevent bloating too, such as

Tip: Ointments with petroleum are flammable and may cause damage to the mask; therefore, petroleum products should not be used as a skin protectant. Oxygen should not be used along with petroleum products, like Vaseline.

those that promote mobility of the digestive system.

Reflux caused by assisted ventilation may be resolved by medication to help alleviate the symptoms, adjusting pressure settings on your machine, or elevating head while sleeping.

Dryness may be resolved by increasing the amount of water you drink during the day. Using a humidifier with your ventilator will moisten the air that you are breathing to help with chapped lips, thicker secretions, and nose bleeds that might occur.

### Invasive Ventilation

Endotracheal tube: consists of a small tube which is inserted through the mouth directly into the windpipe to assist with



### Our Family's Experience

I would like to let you know that our son with myotubular myopathy was discharged from the hospital after four months! We did not consent to the trach despite pushing from doctors, nurses, and other hospital staff. Instead, we asked that he be weaned from oxygen and he did fantastic!

He entered a three-week rehab program at the hospital to regain some of his strength and is now at home with us and his sibling.

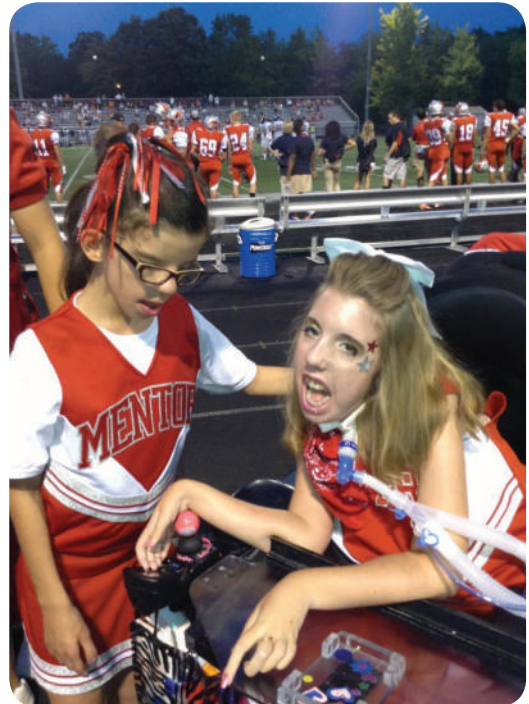
breathing. Usually this form of invasive ventilation is only used temporarily and in emergency situations for someone who is not normally ventilated or who would normally use non-invasive ventilation.

It is more commonly used during the early neonatal stage if the infant with CM experiences severe breathing difficulties associated with their respiratory muscle weakness. It may also be used in older children or adults with CM during any period of hospitalization with severe breathing difficulties which might have been caused by an illness, or as a result of delayed recovery from a surgical procedure.

Tracheostomy: A tracheostomy is a small hole that is made by a surgeon at the base of the throat so that a tube goes directly into your windpipe (trachea). Then, the breathing machine tubing can be directly connected to the tube in the throat.

This may be necessary in people who need long-term mechanical ventilation and for whom non-invasive methods were not successful. Sometimes a tracheostomy is temporary, such as after a surgery or serious illness, and can be removed later.

Throat muscle weakness and all-day breathing assistance needs are two of the most primary reasons for choosing a



tracheostomy over non-invasive ventilation.

Having a breathing tube directly in the trachea is sometimes called a trach, trache, trachy, or tracke depending upon where you live in the world. The decision to have a tracheostomy or not should be considered carefully by your family and the



### Our Child's Trach

Everyone has very different opinions about invasive versus less invasive approaches to

breathing support, but personally we feel that the trach isn't so invasive. Yes, it is very scary to think about but we are so used to it now. Outside of dealing with the issues associated with our child always having to have a parent or nurse who is trained to manage her medical care with her at home or at school, we find the trach to be very manageable and our child leads a very busy and typical life.

### Trach Care Emergency Kit Suggestions

- Same size tracheostomy tube
- Saline
- One size smaller tracheostomy tube
- Water based lubricant
- Suction catheters
- Bronchodilator MDI
- Self-inflating resuscitator bag
- Oxygen, with CO2 and O2 monitor
- Trach ties
- Plan of care
- Hemostats
- Emergency phone list with emergency contacts, physicians, home, cell.
- Suction machine
- Aerochamber
- Scissors
- Manual suction



### Handling Colds and Respiratory Illnesses

During a cold or similar illness, a weak cough can become weaker and it is more difficult to clear mucus from the lungs. Keeping well hydrated is important. People with a muscle disorder have normal lungs, so if the oxygen saturation is going down, it normally means more help is needed to cough and breathe, NOT that extra oxygen is needed.

You may find the following steps helpful in keeping the lungs clear and keeping oxygen saturations above 94%.

1. At least every 4 hours:
  - A. Cough Assist in 5 sets of 5 breaths, ending on inspiration
  - B. Airway Clearance for 10-20 minutes (chest percussions or vest)
2. Use Cough Assist whenever you hear or feel a rattle in chest. Suction mouth when mucus is stuck in back of throat.
3. If you have a breathing machine at home (like BPAP), use it during all times of sleep, including naps.
4. Use the oximeter three times a day when sick and at least once a day when feeling better. If oxygen saturation is less than 94%, use Cough Assist to clear secretions and then recheck oxygen saturation. If oxygen saturation is 92% or less, or if the Cough Assist is not helping to increase the oxygen saturation to at least 94%, then call your respiratory doctor right away.

medical team. A tracheostomy may be a necessary life-saving procedure for some people with CM. See the Appendix for resources on tracheostomy decision-making, online care guides, and networking with other families.

### Sudden Respiratory Illness

Illnesses can be severe in people with CM and can worsen breathing and feeding difficulties. Respiratory infections, including the common cold and flu, are the main causes of hospitalization and should be treated quickly and aggressively.

Upper respiratory tract illnesses develop in the nose, sinuses, ears, and throat. Lower respiratory tract illnesses develop in the lungs. You may need to seek hospital care, such as IV fluids, antibiotics, breathing assistance, or feeding alternatives, to recover from your illness.

Recognizing the signs of sudden respiratory problems is key to finding out if hospitalization is needed. Symptoms include:

- Long-lasting high fever
- Fatigue
- Tired looking or pale



- Appears to be spending all energy just breathing
  - Little appetite and poor eating
  - Increased heart and breathing rate
  - Abnormal movements of chest and belly
  - Weak cough
  - Inability to cough up mucus
  - Chest rattle
  - Oxygen saturation below 95% as measured by a pulse oximeter

If these symptoms are found, call your doctor immediately. When you are being evaluated for your illness, the medical team will do an exam and testing to decide whether or not to hospitalize you. These tests and interventions

may include:

- Determining cough strength
- Pulse oximetry and carbon dioxide measurement
- Chest X-ray to find out if there is a pneumonia or collapsed lung
- Blood and sputum cultures and other blood work
- Respiratory treatments
- Help with breathing (such as BPAP/ventilator)
- Cough assistance devices
- Antibiotics if a bacterial pneumonia is suspected

During illness, the ability to cough and clear mucus worsens.

Without the ability to cough well, mucus and other particles stay in the lungs and create a breeding ground for bacteria and viruses. Breathing problems or a mucus plug can sometimes quickly lead to collapse of part of the lung (atelectasis). It is important to do airway clearance every four to five hours to keep the lungs clear and oxygen saturations at 95% or higher. It is also important to allow your child to rest, repositioning them regularly. Increase their fluid intake if possible, and increase calories as tolerated.

## Aspiration Pneumonia

Aspiration is when something other than air goes into the lungs. Aspiration pneumonia is a lung infection often caused by aspiration of mucus, saliva, food particles, liquids, or stomach contents (reflux or vomit). Symptoms of aspiration could include gagging, attempts at coughing, hoarseness, soreness or burning in the throat, wheezing, chest rattle, and shortness of breath or a halt in breathing. Aspiration also can be “silent,” with no obvious symptoms. Some parents have reported their children’s skin looking ashen or bluish in color with the eyes rolling back during an episode of aspiration. If you suspect aspiration is a problem, immediately call your doctor who may schedule one or more of the following:

- Chest X-ray
- Barium swallow study, also called videofluoroscopic swallowing study
- Visual examination below the back of the throat (laryngoscopy)
- Examination of the airways (bronchoscopy)
- Examination of the esophagus, stomach, and the first part of the small intestine (upper endoscopy)

There are ways to minimize aspiration and subsequent problems that can occur in people with CM. Physical positioning, medications, careful management of oral intake and activity surrounding feedings, feeding tube alternatives, and oral stimulation therapy may help to minimize the risk of aspiration. To reduce the risk of infection when aspiration does occur, ensure good oral hygiene. This will lessen the number of bacteria in saliva and decrease risk of infection.

A common misconception is that artificial airways (tracheostomy) prevent aspiration. Research shows they actually can increase the risk because of reduced elevation

of the air passage around the voice box called the larynx and minimal laryngeal sensation, abnormal laryngeal closure, and loss of protective reflexes often seen in trached patients. Frequent suctioning of ventilated patients is necessary to remove secretions from the nose, mouth, and back of the throat. Maintaining the head of the bed higher, perhaps at 30°, may reduce aspiration in such patients. If they have the strength needed to do so, people with permanent tracheostomies should be taught to remove mucus by coughing, hawking, or spitting (expectorate) frequently and should be instructed to use positioning to avoid aspiration.

More details on how to minimize risk of aspiration can be found in the Feeding, Nutrition, and Oral Care chapter.

## Preventing Breathing Problems

Avoiding illnesses and reducing the severity of infections are critical to preventing breathing issues from worsening.

### Reduce Exposure:

- Thorough hand washing of the affected person and everyone who comes in contact with the person. When soap and water are not close by, use hand sanitizer.
- Stay away from others with symptoms of illness, such as runny nose, coughing, and fever.
- Avoid crowded places like shopping centers, subways, and airports.
- Restrict exposure to groups of children, like in day care centers or schools, during the cold and flu season.
- Avoid eating or touching the face when hands are not clean.
- Stay well hydrated and well nourished.
- Do not share food, drinks, eating utensils, or lip balm with others.

Avoid toxic smoke, such as from tobacco and open fires.

### Vaccines:

- Pneumonia vaccine (pneumococcal vaccine) given at least once after age two
- Flu shots every year (influenza vaccine)
- Routine vaccinations
- Respiratory syncytial virus (RSV) protection is recommended for children under 2 years of age who

are considered high risk for RSV infections. Although the protective drug cannot prevent RSV, it can reduce the severity of symptoms. Check with your health insurance company before starting this monthly treatment, because some will not cover the high cost. Five monthly shots during RSV season, which typically lasts from November through April, are required for protection. If your child is over the age of two and you would like more information about the efficacy of RSV protection, please consult your doctor.

### Cough Assistance:

We cough in order to expel mucus from the lungs, which helps prevent infections. When there is considerable weakness in the neck, rib muscles, diaphragm, and abdomen, coughing ability is impaired, as often occurs in people with CM.

There are two approaches to cough assistance: manual and mechanical.

Manual cough assistance involves a caregiver applying firm and rapid pressure to the upper abdomen and diaphragm, manually assisting in forcing air out of the lungs. This can also be done manually to yourself by folding your arms under your ribcage, leaning forward, and coughing.

A device called a manual resuscitator (trade name Ambu® bag) can be used along with breath stacking to increase lung air volumes before manual chest and abdomen compression as the person with CM exhales. A respiratory therapist or other experienced health care professional can provide training on this maneuver.

Mechanical cough assistance using a mechanical insufflation-exsufflation device mimics a natural cough. The machine gradually delivers a large volume of air through a tube attached to a mask that covers the nose and mouth, or a tube can be attached to a person's trach. This incoming air is just like when you breathe in (positive pressure). Once the lungs have been expanded (similar to a normal deep breath before a cough), the device quickly reverses the flow of air to pull secretions out of the lungs (negative pressure). Mechanical cough assistance helps to make your cough stronger and more effective. This helps keep your airways clear to reduce the chance of recurring respiratory infections.



Cough assistance techniques can also be used as therapy for your breathing muscles to help keep them stretched and flexible. It is more effective in children over the age of 2 years, but has been used in infants with more severe forms of CM to good effect.

### Assistance with Clearing Mucus:

There are many ways to help keep the lungs clear. If you need help learning the techniques or using the tools, contact your respiratory therapist for instructions to ensure you and your family members are confident when it has to be done at home.

- Breath/Air Stacking—A manual technique that uses a bag or breath holding and stacking to help push more air into lungs to prevent collapse of lungs (atelectasis).
- Frog Breathing—Called glossopharyngeal breathing by doctors, this technique involves use of the tongue and throat to force extra air into the lungs.
- Manual Chest Percussion—Gently hitting the back (percussing), using either your cupped hand or a cupped tool, to loosen mucus so that it can be removed. This may be done when ill or regularly when feeling well.
- Postural Drainage—The person is positioned at an incline with head and chest lower than the hips. This can be achieved using pillows or a wedge. Gravity helps pull mucus from the lower lungs into the upper airway, allowing them to be removed through suctioning or coughing. This technique, however, may not be appropriate in someone with risk for aspiration.
- High Frequency Chest Wall Oscillation Vest (Percussive Vest)—A vest that is worn and attached to a machine that vibrates the chest and helps loosen mucus, which is then

coughed up or removed with a suction device. Discuss vest use with your clinicians, as it may not be appropriate for everyone.

- **Intrapulmonary Percussive Ventilation (IPV)**—A mist of saline and burst of gas are delivered throughout the entire respiratory cycle using this machine that loosens and mobilizes secretions in the lungs. Use with a mouthpiece or inline for trach and vent users.
- Medications to break up mucus or open up airways can also be used when sick, such as albuterol (salbutamol) or dornase alfa. A machine called a **nebulizer** delivers these medications as a mist to the lungs through a mouth piece, mask, or inline for trach and vent users.
- Saline mist delivered through a nebulizer helps to moisten, thin, and loosen secretions, making them easier to remove.
- Good hydration keeps secretions thin and easier to remove, so make sure you are getting plenty of water and other liquids in your diet daily.
- **Suction Machine**—A powered device that has a thin, plastic tube designed to suck up mucus and excess saliva, usually from the nose and mouth or trach. This is to prevent the secretions from lodging in the wind pipe, which can result in aspiration pneumonia or sudden blockage of the airway, which is a breathing emergency.

### Managing Excessive Oral Secretions

Some people with CM have too much saliva that cannot be swallowed, therefore it spills out of the mouth as drool or may be aspirated into the lungs. This can be caused by having overactive salivary glands, an uncoordinated swallow, and/or poor lip closure.

Some treatments for excessive saliva include:

- **Speech Therapy**—to improve jaw stability and closure, increase tongue strength, and improve lip closure; best started in infancy for optimal results.
- **L-Tyrosine**—an amino acid found in health food stores and at pharmaceutical suppliers that has been found to reduce secretions in some people. No doctor's prescription is needed to obtain this dietary supplement, but do let your doctors know if you are taking it. There are side effects and it should be discussed with your doctor prior to use.

- **Glycopyrolate**—a medication dosed at 20-100 micrograms per kg for each dose given every 6-8 hours.
- **Scopolamine Transdermal Patch**—each patch is 1.5 mg and is changed every 2-3 days. Some patients may benefit from a lower dose due to the potential for thicker secretions or mucus plugs.
- **BOTOX®**—can be injected into the salivary glands under ultrasound guidance to reduce the amount of saliva produced.
- **Salivary Gland Ligation**—surgery to close off some of the saliva glands.

The least invasive treatments for managing oral secretions should be tried first, followed by surgical treatment.

### Breathing Exercises

Exercises to help the lungs fully inflate will improve development of the lungs and can increase muscle strength and function.

Things you can do without any equipment include breath stacking, stretching and massage of the intercostal muscles, and deep breath holding for several seconds. Singing or blowing into a harmonica are fun activities for children that can exercise breathing muscles. You can also use a mechanical cough assistance machine or ventilator as a therapy tool.

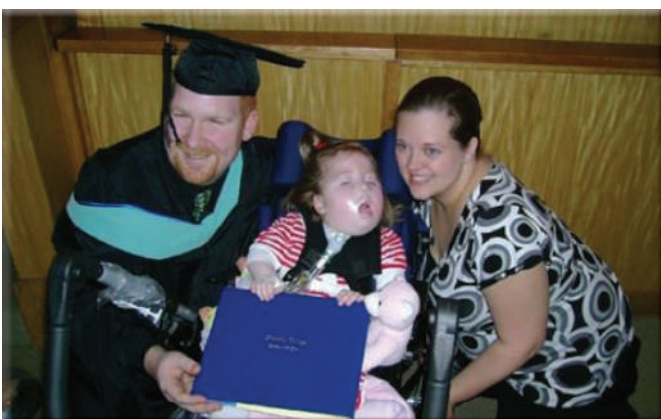
Development of the lungs in infants and young children is important. Body movement and crying are a couple of ways that the respiratory system can get a workout naturally. But, when weakness is severe, you may need to find creative ways of encouraging movement and use assistive devices to help give the lungs a workout. These are described in the Appendix.

### Emergency CPR

It is recommended that everyone involved in the care of someone with CM be familiar with emergency resuscitation, called **cardiopulmonary resuscitation**, or CPR. Classes are offered through hospitals, clinics, schools, and organizations, often at little or no cost. You can also find tutorials on the Internet to learn basic procedures that could help keep your loved one breathing until the emergency medical team can arrive.



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 A manual resuscitator (known by the trade name Ambu® bag) can be used for air stacking or lung inflation/range of motion, temporary breathing assistance, or in breathing emergencies. It can be used with a mouth piece, mask, or trach.  
 .....



### Appendix Items

- Cough Assist Protocol, Adult
- Extubation Plan
- Respiratory Emergency Letter
- Recommendations for Post-Operative Care and Extubation of Children and Adults with Neuromuscular Disease
- How can I encourage movement to help development of the body and respiratory system?



# 4

## Orthopedics & Rehabilitation

The branch of medicine that deals with the prevention or correction of injuries and issues of the bones, muscles, joints, and ligaments is known as orthopedics. Muscles control the position and movement of the bones, so it makes sense that muscle weakness would likely lead to problems with the bones of the body. Orthopedic problems involving the skeletal system are frequently encountered in congenital myopathy (CM) and include the following:

- Difficulty walking
- Scoliosis
- Hip joint out of place (dislocation or subluxation)
- Foot and ankle deformities (such as clubfoot)
- Joint tightness (contractures) including a severe form called arthrogryposis
- Weak or thin bones (osteopenia or osteoporosis)
- Pain or discomfort

Your orthopedic and rehabilitation specialists will play an important role in assessing these issues. Many interventions are done ahead of time to prevent permanent limb changes (deformity) or respiratory complications in the future.

The goals of orthopedic and rehabilitation care are to:

- Keep you flexible and mobile
- Improve strength where possible
- Maintain the best posture for your situation
- Prevent or delay joint deformity
- Keep your bones as strong as possible
- Minimize pain and promote comfort
- Provide devices to help you be more independent

Although all subtypes of CM can include orthopedic complications, the following three genes are related to them

more often than not:

- *RYR1*
- *SEPN1*
- *MTM1*

### How am I evaluated for orthopedic and rehabilitation needs?

The rehabilitation specialists—which include a physical medicine and rehabilitation doctor, occupational therapist, physical therapist, orthotist, and wheelchair or other equipment specialist—will evaluate your joints, spine, sitting comfort, mobility, and your ability to do your daily activities. They will evaluate you by doing a physical examination, and may use a tool to measure muscle strength (myometer), order breathing tests, or X-rays.

The orthopedic surgeon will evaluate the need for surgery, such as to treat scoliosis. You should be seen by a rehabilitation specialist at least annually and an orthopedic surgeon at regular intervals when it becomes necessary.



## Scoliosis Management

Scoliosis is a side-to-side curve of the spine, while kyphosis is the backward bending of the spine, and lordosis forward bending. Any curve more than 10 degrees is considered abnormal. These spinal deformities can cause difficulty walking or sitting, pain, and if severe, can worsen breathing ability. It is a common orthopedic problem in CM. Once found, an orthopedic surgeon that specializes in neuromuscular disorders should be contacted.

There are several different signs that a patient or parent can be aware of themselves, to help determine whether they or their child actually have scoliosis. If one or more of the following signs are present, it is advisable to schedule an exam with your doctor.

- Shoulders are different heights
- One shoulder blade is more prominent than the other
- Head is not centered directly above the pelvis



- Appearance of a raised, prominent hip
- Rib cages are at different heights
- Uneven waist (more prominent crease on one side)
- Changes in look or texture of skin overlying the spine (dimples, hairy patches, color change)
- Leaning of entire body to one side and loss of balance when sitting

If your local hospital or clinic does not have someone with expertise in treating patients with CM, you should look outside of your hospital system. Connecting with other affected families in your region may help you find the clinical expertise you need.

**Sitting**—Children aged 18 to 24 months who are not yet sitting may benefit from temporarily using a soft spinal brace for short periods during the day.

**Leg length**—For those that are ambulatory and have one leg shorter than the other (leg length discrepancy), this will cause the pelvis to dip down on one side. Then the spine will curve

### Intervention Stages for Scoliosis

Before it starts:

- Check for unevenness (asymmetry) and correct it
- Stretch and strengthen the core muscles daily
- Have good posture when sitting, standing, walking, and sleeping
- Get supportive seating and shoes

Once it is first noticed or diagnosed:

- See a specialist to learn targeted stretching and strengthening exercises
- Get evaluated for a rigid brace
- Increase your level of activity to stay flexible and as strong as possible
- Carefully monitor breathing ability as it may change at this time

Once it has progressed to 30-50 degrees:

- See an orthopedic surgeon highly experienced with correcting scoliosis in people with CM for guidance on your options
- Maintain healthy diet and stay active to be in best condition for possible surgery

as an adaptive response, leading to an abnormal, fixed curve. Have a shoe insert made to correct the difference in leg lengths and wear it in your shoe for all standing and walking activities.

**Seating**—Weak trunk muscles (along the spine, abdominal, shoulder, and pelvic muscles) lead to a floppy or slumped posture. Children and adults with CM might bend their backs into a position where they can find support, leading to greater curve of the spine. Seating that promotes a symmetrical, upright posture and offers support for when the person fatigues is critical.

**Degree of curvature**—When scoliosis is found, the doctor will likely do X-rays approximately every 4 to 6 months to monitor the curve. Doctors measure the curve amount by degrees. The higher the degree, the worse the curve:

Any curve of the spine between 10 to 24 degrees should be

carefully monitored by you and your doctor. It is recommended that your stretching and core strengthening activities increase in frequency at this time.

- A curve between 25 and 50 degrees should be treated with strategies like bracing and physical therapy programs.
- Curves over 50 degrees often require surgical correction. The better your condition and health prior to surgery, the more promising your outcome is likely to be. There are individual and regional differences for when scoliosis surgery may be recommended.
- It is important to do regular stretching and massage of tight, contracted spinal muscles, along with strengthening over-stretched muscles.



## Coming to Terms with My Scoliosis

As a teenager, I was told by my doctor that I have scoliosis. This news, combined with my muscle disorder, was quite a blow to me. I felt defeated because I thought I had failed to keep my core strong enough. I feared that my body would become so distorted by the curving of my spine that other people would shrink away from me. My future seemed to have turned bleak in a flash. I felt alone and depressed.

I had to get fitted for a Boston brace, which is a type of rigid brace. The measuring process took half the day and I needed to wear a body stocking. It took about two months for them to make my brace and then I had to go back for half a day to get it fitted.

There are a lot of different styles of braces, but mine is tan colored and opens in the back. There are locking straps that hold it very tightly onto my body, which can make it hard to breathe sometimes. I have to remove the brace in order to drink, eat, use the bathroom, or walk very far. I try to wear my brace during sedentary activities, such as whenever I am sitting at my computer, watching a movie, or reading a book. I try to stay active, so I do not wear my brace if I am engaging in movement. The rigidity of the brace makes it too hard for me to move around much while wearing it.

I try to exercise every day, no matter how much I don't want to do it. I have seen proof in my own body that exercise really helps. Now I'm not saying I'm a muscle woman, but I can promise you, I am a lot better off now than I would have been if I didn't exercise. I perform exercises to strengthen my core, which makes the muscles around my spine stronger. I think this helps the most in slowing or stabilizing the curve of my spine.

Although I had to make some changes in my life and the way I think, I was able to make it through this challenge relatively intact. This is a part of my life now, and although it is hard, I've come to accept that.



## Bracing

Some doctors recommend bracing as a part of treatment while others do not. If your doctor recommends a brace, it should be customized to allow for maximum lung expansion while wearing. The specialist may place a cut-out where the diaphragm expands the torso outward.

Pulmonary function tests could be performed in the brace and out of the brace to determine if the brace is limiting your breathing ability. A rigid brace will act as a total support that holds the torso up with virtually no muscle activation and can lead to worsening muscle weakness (atrophy). If you are concerned about a rigid brace leading to muscle atrophy, it may be best to wear the rigid brace only during sedentary tasks, like reading or watching television.

Some rigid braces are not worn during sleep due to the pressure the brace imposes upon breathing muscles. If you use a ventilator it might be possible to wear your brace during sleep, but check with an experienced medical professional before wearing it at night or during naps.

For those with severe core weakness, bracing may be the only way for them to sit upright and can be a good option to improve sitting posture.

## Surgery

The goals of scoliosis surgery are to preserve lung function, mobility, and posture for as long as possible. There are many types of surgeries and a recommendation will be based on your particular situation. In general, your medical team will try to treat scoliosis using bracing and other conservative methods for as long as possible. Treatment will vary by geographic location.

Surgery using “growing rods” will often be used if a child is still growing and less than 10 years old. The rods are inserted into the spinal column and can be lengthened through a small

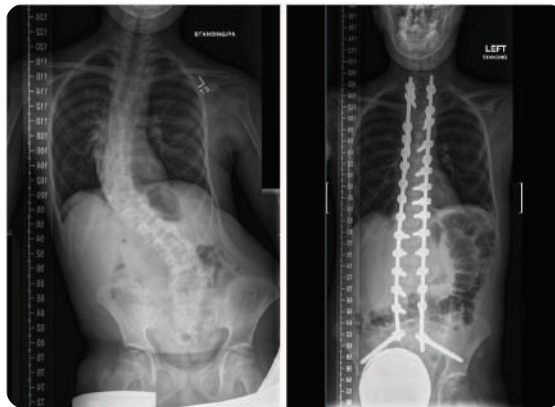
surgery every year or so to allow a child to grow in height. This option does carry some extra risk since it requires multiple smaller spinal surgeries. When a child has finished growing, a final complete “fusion” surgery is performed. Future treatments may include magnetic growing rods which minimize invasive procedures to extend the rods as the patient grows.

Surgery is not for everyone. You will have to make a decision after a full discussion with your medical team, family, and friends from your neuromuscular community. One of the most important reasons to choose scoliosis surgery is when the curve has altered the shape of the rib cage and diaphragm to such an extent that breathing is compromised. Another

reason to have scoliosis surgery is to help with pain caused by the curve. Due to worsened weakness following the surgery, some patients who could walk and breathe on their own before surgery lose that independence following scoliosis surgery.

Spinal fusion surgery may not completely correct the curve depending on the severity and stiffness of the spine at the time of surgery. Even after you are done growing (skeletal maturity),

the spinal deformity can slowly get worse without surgery over time because of muscle weakness.



## What to expect if you must undergo spinal surgery

### Before Surgery:

- An evaluation with a dietician for optimal nutrition several months prior to surgery is recommended to make sure you are in the best condition and health.
- An evaluation of your jaw and neck range of motion as well as an assessment of your heart and lung function.
- Breathing measurements (pulmonary function tests) are necessary. Your lungs need to be strong enough to withstand surgery.
- Make an extubation plan with your respiratory doctors.
- Take baseline videos/photos (using your smartphone) of your function, so that the doctors can see when your normal function has returned (i.e. walking, breathing, sitting).

- Anesthesia should be carefully chosen because some mutations, especially those affecting the RYR1 gene, are known to cause **malignant hyperthermia (MH)**. The muscle relaxant succinylcholine and anesthetic agent halothane should be avoided. The anesthesiologist should use a “clean technique,” which means that the equipment was cleaned of all traces of anesthesia used on a previous patient. They should also have the MH rescue medication, dantrolene, on hand in the surgery room. Make sure your surgery team knows you may be at risk for MH.

#### After Surgery:

- You will need aggressive and expert pulmonary care.
- You will be cared for in the Intensive Care Unit.
- You may be on a mechanical ventilator to assist your breathing for a while, even if you have never needed to before. Your doctors will “wean” you off the machine gradually if mechanical ventilation is not needed long-term.
- Airway clearance may be helpful post-surgery.

It is common to be weaker after a major surgery, so aggressive rehabilitation is very important. By getting you mobile soon after surgery, your therapist can help ensure your weakness does not worsen.

Rehabilitation therapists will evaluate you for the following:

- Feeding—self-feeding may be difficult, requiring use of adaptive devices or a temporary feeding tube to help you eat
- Mobility—transfers, wheelchair modifications, and home care support to regain your strength and range of motion
- Head and neck strength—you may need support after surgery
- Pain—your pain should be addressed immediately in the hospital and with continuing care once you are back home
- Constipation — constipation can worsen after surgery and should be addressed early-on
- Scoliosis and lung health—Continued monitoring of scoliosis as well as your lung function after surgery

## Hip Dislocation

Hip dislocation (subluxation of the hip) occurs commonly in CM

at birth or anytime during infancy, especially with central core myopathies due to RYR1 mutations.

Preferred treatment is splinting with a special brace called a Pavlik harness until the joint becomes tighter. This brace helps position the hip properly. The hip may be immobilized for a few weeks. Immobility for more than 12 weeks is not recommended because contractures may develop.

If a child is not walking, then surgery is typically not done. Surgery may be done if the dislocation causes severe pain (very rare) or if it is worsening sitting ability or scoliosis.

If a child is walking, then surgery needs to be considered very carefully, as it can worsen weakness and may even result in the loss of walking ability.

## Knee Contractures

Most of the time, no surgery is needed for knee contractures. Surgery can be considered if it stops you from sitting in your wheelchair comfortably.

## Ankle Contractures

Newborn babies with clubfoot should have non-surgical correction using serial casting or nighttime splinting.

Serial casting is when several casts are used, one after another, until the desired position is achieved. Each cast can be positioned a little further than the last, allowing the goal to be reached slowly over time.

Splinting involves wearing a rigid or flexible device that covers the bottom of the foot and half-way up the lower leg to keep it in a specific position during sleep.

Heel cord lengthening surgery can also be done. An older child or adult with foot deformities will need to carefully weigh the surgery’s risks and benefits against the possibility of increased deformity and pain.

## Leg Bone Fractures

A fracture of your leg bone, no matter the cause, should be treated with splinting, casting, or surgery followed by

aggressive rehabilitation to prevent loss of ambulation, if achieved.

Surgery may involve inserting a metal rod into the bone to stabilize it. Rehabilitation should start soon after surgery.

Casts should be lightweight and can be designed to include “joints” that still allow for walking.

## Bone Health

Bones can easily become thin (osteopenia) and brittle (osteoporosis) as a result of limited mobility and weakness. This makes the bone prone to fracturing, sometimes even without trauma.

The following can optimize bone health:

- Weight-bearing activity, including movement, exercise, and even standing
- Make sure diet includes the recommended daily intake of calcium; if needed, supplement with vitamins
- Make sure diet includes the recommended daily intake of Vitamin D (cholecalciferole) and get enough healthy sun exposure; if needed, supplement with a vitamin. Vitamin D levels can easily be checked with routine blood work.

If a person with CM experiences a fracture, their doctor will most likely monitor bone density through a scan called a DEXA scan (dual energy X-ray absorptiometry).

## Treatment of Very Brittle Bones

In some cases, medications may be used to help people with severely brittle bones. These types of medications are called bisphosphonates and should only be administered by a doctor with experience using them in muscle disease.

Bisphosphonates should be considered when a person with CM has:

- Two or more fractures
- Bone pain
- Severe osteoporosis as seen on regular X-rays
- Failure of pins and screws from bone surgery due to weak bones
- Other health conditions that worsen bone health (e.g. celiac disease, thyroid disorders, and delayed puberty.)

## Maintaining and Increasing Mobility While Preventing Skeletal Problems

Being proactive about your care is the best way to help prevent, minimize, or at least delay skeletal problems.

### Exercise

Staying active can help maintain and improve your muscle strength and function. A physical or occupational therapist may recommend exercises, stretches, and/or splinting, based on your needs. In addition to the therapies prescribed for you, exercising on your own is important to see and maintain improvement.



**Aerobic Training**—To the extent you are able, do exercises that involve your entire body and use up oxygen, like walking and swimming. This is commonly referred to as “aerobic” exercise, and it can be done three to five times a week without risk. This means you should do as much as you are able to do, but never to the point of exhaustion.

### Signs You May be Over-Exercising

1. Pain or cramping in muscles
2. You feel weaker a day or two after exercise
3. Heart rate above 150 beats per minute
4. Shortness of breath

For those with significant movement limitations, please seek advice from your physical therapist. Not all people with CM will be able to perform aerobic training.

**Resistance Training**—Do lifting activities using your own body weight, resistance bands, machines, or light weights, to the extent you are able. Resistance training may increase bone density and bone strength, in addition to potentially improving muscle strength. Methods include:

- Using gravity alone to impose resistance through lifting your arms up as high as you can. This is enough to sufficiently exercise the muscles for some people.
- Using elastic bands or tubes that stretch when pulled. These provide more resistance than gravity alone and therefore will challenge muscles more.
- Using machines, such as a leg press or stationary bike, commonly found at gyms or available for purchase.
- Light free-weight lifting is sometimes recommended beginning in the late teens for those who are able. Weights should be lifted with good technique and under direct supervision (with a “spotter”) to prevent injury. Your goal should not be to “bulk up,” but to make some gains, such as in strength, function, or mental wellness.

If you have decreased range of motion, then your medical team should help guide how much, if any, resistance training you should do. Resistance training that feels painful or leads to overexertion is not advised.

**Balance/Core Training**—If able, perform exercises that require balance, such as sitting unsupported, sitting on a large exercise ball, or standing on a wobble board with supervision.



Where available, therapeutic horse riding (hippotherapy) is another balancing activity that promotes core muscle strengthening. Keeping your core as strong as possible may help prevent or postpone curving

of the spine, as discussed in this chapter.

All exercise programs should be started slowly, with gradual increases and moderation in mind so that you are not terribly sore or severely fatigued for days after the exercise. It is acceptable to feel a little sore or tired for a day following a physical challenge, like exercise will induce. Stretching before and after exercise can limit muscle soreness and prevent stiffness or injury.

Consume plenty of fluids throughout your exercise and after your routine. Hydration and good nutrition are essential to maximize your performance outcome and help prevent muscle cramps.

Motion in warm water is a great way to promote movement for most CM subtypes.

Never exercise to the point of exhaustion.



For sports lovers, high impact sports that could lead to injuries should be avoided. Please ask your medical team if your selected sport could put you at risk for harm before undertaking this activity. If your CM could have associated heart complications or metabolic dysfunction, discuss exercise options and risks with your medical team before exercising.

## Standing

Encourage very weak children who have not yet started to stand on their own—and others who do not walk but can stand—to stand several times daily. Standing frames, parapodiums, tilt tables, and orthotics can help maintain upright posture.

Standing helps to:

- Minimize stiffness and locked joints (contractures) in legs
- Develop trunk, head, and pelvic stability
- Exercise muscles used for standing
- Promote independence and self-esteem
- Strengthen bones

Care must be taken to make sure that the equipment straps are not too tight, especially across the knees, because it may cause fractures if bones are very thin. Your physical therapist or rehabilitation specialist will determine how long and how often to stand based on your situation.

### Stretching and Range of Motion Exercises

Stretching various joints on your own (active stretching) and by someone else (passive stretching) helps maintain the amount of movement typical of a joint (range of motion) and prevents or delays contractures. For those with more severe CM forms, stretching may be one of the few exercises possible to perform.

Joints that commonly need stretching include ankles, hips, knees, neck, spine, elbows, wrists, and fingers. Your rehabilitation specialists will evaluate you to see if you need an orthosis, which is a device used to support joints.

People who spend most of the day sitting can develop tightness of the ankles, knees, and hips, which can be prevented or delayed through stretching. For those who are ambulatory, maintaining ankle flexibility is critical to continue walking ability as long as possible. Following are some things you can do that usually help.

Ankles—These interventions help maintain walking and independence:

- Daily stretching of ankles by family member or yourself
- Wearing night-time ankle-foot orthoses (AFO)
- Wearing day-time dynamic AFO, molded AFO, or knee-ankle-foot orthosis (KAFO)
- Standing in place during the day with or without an orthotic
- Progressive splinting—using a soft orthotic device to gradually help stretch the Achilles tendon
- Serial casting—using a hard fiberglass cast to gradually help stretch the Achilles tendon

Knees, Hips—To help maintain a comfortable sitting position in a chair, upright posture, and mobility:

- Daily stretching of knees by family member or yourself by lying down with legs stretched out
- Stretch out hips by lying on stomach if possible
- Wearing AFOs as described above
- Standing in place with a stander or other device
- Progressive splinting
- Serial casting

Shoulder, elbow, wrists and hand—To help maintain hand and upper body function and independence:

- Stretch all joints in upper body several times a day
- Wrist, hand, and elbow splints given by occupational therapy

#### Potential Benefits of Exercise in CM

1. Improves circulation
2. Preserves flexibility and range of motion
3. Maintains or improves physical function
4. Improves respiratory efficiency
5. Upholds overall fitness
6. Sustains or improves bone density
7. Retains or increases strength
8. Helps relieve stress and tension
9. Improves mood
10. Results in better sleep
11. Reduces risk of developing other conditions like diabetes

Spine—To minimize scoliosis, consider:

- Stretches and exercises that use back muscles
- Torso (trunk) bracing in the sitting or standing positions or both

Rehabilitation specialists often use Botulinum toxin (Botox®) to help treat muscle tightness such as in cases of cerebral palsy. However, Botox is not advised for use in the muscle of people with CM because it may worsen the weakness already present. Botox has been successfully used to treat excessive saliva without affecting muscle in some people with CM. Please refer to the Respiratory Management chapter for more information about Botox use in controlling excessive saliva.

**Adaptive Equipment**

One of the main goals when treating CM is to safely help maintain overall mobility and independence. People with CM are at a greater risk of falling and may suffer more severe injuries when they do fall due to an inability to “break their fall.”

For children, not being able to access their environments can lead to secondary cognitive and social dysfunction. Improving mobility can result in many advantages for children, including increased awareness of their environments, better coordination, more vocalizations, and more contact with others, with an enhanced motivation to explore their surroundings.

Use of devices, scooters, wheelchairs, and accommodations can help, such as:

Ride-on toy cars for children’s mobility—These can be modified to suit individual needs.

Canes, walking frames, orthotics, swivel walkers—These can be used early on to help promote and maintain walking.

Manual and power wheelchairs—Can be used by children as young as 2 years. Points to consider:

- Good support to pelvis, trunk, and head if needed
- Lap belts for safety
- Some may need chest straps
- Comfortable and customized seating to handle all your needs
- It may take some time to receive your power wheelchair, so it’s best to start the process early

Mobility Scooters—Can be used by children and adults who require less seating support and have use of their arms and hands to safely operate the drive mechanism and steering. Points to consider:

- Can be ordered with smaller seats and lap belts.
- Locking swivel seat and adjustable armrests make getting in and out easier.
- Simple to operate.
- Most include a basket in front for belongings and

- accessory storage in back for the power cord.
- Includes adjustable tiller and handle bar steering that requires the user to hold the accelerator with one finger during driving.
- Easily adjust your speed on a dial.
- Can be transported in the trunk of a car or can be driven into a ramped vehicle.
- A good option for those who need mobility assistance sometimes and for getting around outside the home (your community, malls, zoos, museums, etc.).
- Less adaptable for those with special seating needs such as significant arm weakness, or pelvic rotation and back support needs.
- Sitting up without the ability to recline during scooter use can be fatiguing for some people with CM.
- Has a platform foot rest but no elevating leg rests or head supports.



Adapted Vehicles—Once you are of driving age, driver educational programs and modifications to your vehicle may be necessary.

Environmental Modifications (E-Mods)—Changes to your home, work place, or school could include ramps, rails, and stair lift systems.

Parking—Obtain a handicap/disability accessible parking pass. Ask where the most

convenient parking is located for someone with a disability when you are not sure.

Accommodations—Request accommodations such as sitting near the doorway for ease in coming and going from your engagements.

Other—Additional pieces of equipment and aids include a shower chair, reacher, Hoyer lift, transfer equipment, service dogs (canine assistants), and aids to help with eating, drinking, communicating, and toileting.

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**BOTOX® may be used in salivary glands, but should NOT be used in skeletal muscles in those with CM.**  
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### Appendix Items

- Tips for Managing a Scoliosis Surgery



## Feeding, Nutrition, & Oral Care

Feeding and swallowing difficulties are common in CM and can result in poor nutritional status and other health problems. Some children and adults are not able to eat safely by mouth due to swallowing problems either early in life or later in life. Maintaining good nutrition along with gut and dental health is important because it improves growth, energy level, amount of activity, and breathing ability.

The medical team members who will care for this aspect of your health include a speech/swallow/feeding therapist, gastroenterologist, neuromuscular specialist, pulmonologist, occupational therapist, and a nutritionist or dietician. Ideally, they should have experience treating people with CM.

### Swallowing and Nutrition

The best way to monitor your nutritional status is to measure height and weight at every clinic visit for children and adults, and at least every three months in infants. A person that is unable to stand or has scoliosis can have a measurement of the forearm (ulna length) used instead of height.

You can track growth trends in children (height/weight) using a chart called a growth curve on which measurements of each height/weight are plotted on a graph. This allows you and your doctor to see trends over time. Ask your doctor to provide a graph for you.

Children with CM often have growth curves that are below what one expects for age in unaffected children; however all children are expected to maintain their own growth curve. Don't worry as long as your child can maintain his/her level of growth and not "fall off the curve," is not sick frequently,

and is not having recurrent lung or heart problems. Weight percentiles for a child that fall below past patterns would be an indicator that they are not getting enough calories. Infants who are not gaining enough weight are considered undernourished and sometimes said to have "failure to thrive." They are usually referred for special care to get their weight up.

Sometimes people with CM put on too much weight because they are consuming more calories than needed for their activity levels. The extra weight makes it much harder to move around, be transferred, and puts additional stress on the body and organs. Maintaining an ideal weight is important and your doctor can help you to know what that is for you.

### Signs of Feeding Problems:

- Low appetite and food intake
- Chewing incoordination or taking a long time to chew
- Coughing, choking, gagging, gasping, or difficulty



Image credit - <http://www.arktherapeutic.com/post/1266>

- breathing during meal times
- Excessive secretions pooling, drooling, or problems



- coordinating the muscles
- A raspy sound to the voice or to breathing after swallowing or after the meal
- Unable to clear the mouth of food and drink after the swallow
- Food or drink lingering in the throat
- Making throat clearing attempts and noises during meals
- Nasal or respiratory congestion, increased work of breathing, fatigue
- Long meal times; such as much longer than 30 minutes
- Anxiety in anticipation of meals or discomfort during meals
- Excessive spitting up or reflux
- Vomiting or appearing to be in pain during or after eating; a person may complain of chest or abdominal pain after eating
- Delayed advancement to new food textures
- Difficulty advancing to use of age-appropriate meal utensils
- Decreased growth or weight
- Lung infections

### Feeding/Swallowing Evaluation

A speech-language pathologist who specializes in feeding and swallowing can evaluate swallowing ability if you have concerns, but in some facilities this is done by an occupational therapist.

Initially, they will do an exam to assess oral skills and feeding practices. They will watch how you swallow, inspect your mouth, and try different foods and liquids. One of the goals of a feeding/swallowing evaluation is to determine risk for swallowing problems and aspiration. The clinician will try to find out which foods are safe and appropriate for your skill level and decide if further examination is needed.

The exam may involve:

- Discussion about feeding history and current feeding practices
- Oral examination to assess for any structural problems related to eating
- Assessment of strength and coordination of mouth and swallowing muscles, head control, and maintaining posture

- Observation of a typical meal to note oral skills and swallowing
- Trials of different foods and feeding methods to help with efficiency and safety

People may have difficulties with eating because they are lacking oral skills needed to eat. This does not necessarily mean that they have risk for aspiration. However, if indicators for swallowing problems were identified during the feeding observation exam such as coughing, throat clearing, respiratory congestion, changes in vocal quality, and breathing problems during eating, then conducting a video swallow study might be suggested as the next step.

### Video Fluoroscopic Swallow Study

A video fluoroscopic swallow study should be performed whenever swallowing problems are suspected. This test uses different types of food mixed with barium. A series of video X-rays are done as you swallow. Barium is not harmful and will allow your speech/feeding clinician to see the food as it travels from the mouth into the stomach.

Your specialist should be able to identify any problems with strength, coordination, and efficiency. The test will also show if other problems are occurring, such as food getting stuck or going the wrong way. For example, sometimes food can be pulled into nasal passages or into the windpipe (trachea).

If any swallowing problems are seen on the video of your swallow, your specialist will help you decide the next step – perhaps suggesting special feeding techniques or using different types of foods. Some children and adults will need alternative methods of feeding and some may need therapy to help improve swallowing ability over time.

### Chest X-ray

A chest X-ray may be done for evidence of aspiration pneumonia.



Image credit - [http://www.refluxrebels.com/Acid\\_reflux/Acid\\_reflux\\_tests.html](http://www.refluxrebels.com/Acid_reflux/Acid_reflux_tests.html)

## Improving Feeding and Growth

Learning what is most suitable for your feeding needs, understanding the techniques involved in safe eating/feeding, and keeping track of growth and weight over time are important to staying well-nourished. For ideas on how to increase your calorie intake, please see the Appendix.

## Common Feeding Tubes (Nasogastric Tube and Gastrostomy Tube)

For those with the severe neonatal form of CM, having a safe method of feeding will need to be decided right after birth.

For those with moderate or mild CM, feeding by mouth may be feasible, possibly with some challenges. Not getting enough nutrition by mouth or repeated lung infections due to aspiration can slow down growth and cause health problems. If poor growth continues or other feeding-related health concerns persist despite good effort and guidance from a feeding specialist, then a feeding tube will need to be placed.

The two most common feeding tubes are the nasogastric tube (NG-tube) and a gastrostomy tube (g-tube or PEG tube). Some children and adults who use feeding tubes are also able to eat by mouth. The tube is often used to supplement calories and nutrition that some are unable to consume on their own.

## NG-Tube and G-Tube

Often used for short-term feeding, an NG-tube is a tube that goes into the stomach from the nose. NG tubes are usually placed while you are awake.

A g-tube or PEG tube is a long-term feeding solution. This

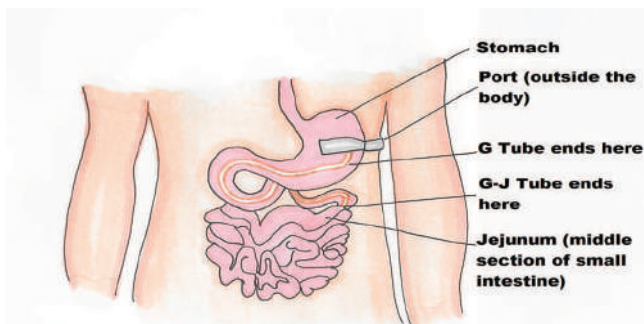


Illustration by Lucinda Ladwig.

tube goes directly into the stomach through the skin. G-tubes and PEG tubes are typically placed while the patient is under anesthesia by a GI doctor. If the risk of anesthesia and respiratory problems are significant, you may want to ask about other options.

Liquid food (commercial formula or blenderized diet) can be given through the tube directly into the stomach, so that people with CM can safely get enough calories to grow and stay healthy. Your dietician will be an important resource for gauging how much to feed, the calorie content of meals, the frequency of feeding (bolus or continuous feed), and what nutritional supplements will be needed.

A long-term feeding solution is highly recommended in newborns with CM who are very weak. See the Appendix for a complete list of feeding tube options along with some pros and cons of each feeding method.

## Oral Stimulation

It is important to maintain positive oral experiences for your child. Children who are not allowed to eat by mouth or those that have negative experiences every time they eat may develop an oral aversion. This is a powerful dislike and firm avoidance of food being in or around the mouth and can occur in babies and children that are tube fed.

Your speech-language pathologist or feeding specialist can give you some ideas of what to do, such as mouth-play during tube feedings, so that your child will accept food by mouth when ready. A clinical psychologist may also be helpful. Your therapist can also suggest ways to help strengthen and

.....  
 If your child needs a g-tube, you can create tube feeding meals made from a great variety of foods through homemade blended formula. This can be as simple as adding jarred baby food to commercial formula or can consist of a diet composed of only blended foods. To get started, find a registered dietician with experience in blended formulas and see the Appendix for resources.  
 .....



## How One Mom Transitioned her Child From G-Tube to Oral Feeds

My daughter was born with congenital myopathy and had feeding problems from birth necessitating a g-tube for all nutrition. I anticipated that through therapy, her swallowing function would improve and I wanted her to have the option of oral feeding once she was ready. This story describes what worked for us to transition my daughter to oral feeds. When planning to transition from tube to oral feeds, daily oral stimulation activities will be key to combatting oral aversion. This is just our experience, shared in the hopes that it may be helpful to others. Your clinician may have other suggestions for you.

### Before Mealtime:

We played with sensory-stimulating toys around my daughter's cheeks, chin, lips, neck, and fingers. These included koosh and nubby balls, bubbles, massagers, vibrating toys, sensory teethingers (warm/cold), and other toys or books with interesting textures. I kept these toys only for use as pre-feeding activities.

### Introducing Taste and Texture During Mealtime:

I applied tastes of food to my finger, pacifier, or oral brush and placed the food on her lip or tip of tongue and encouraged her to taste it. I began with mild, diluted tastes and slowly increased taste intensity according to her responses. I sometimes dipped a clean baby cloth into water for her to suck on. Small drops of flavored liquid offer the least texture and are easier to start with. I varied the taste gradually (not just sweet) so that she would accept the taste of many foods from all food groups, including vegetables. Children are drawn to foods given at an early age and I wanted preferences to include healthy foods.

My daughter needed to follow bites of purees with liquid to help clear the back of the tongue and throat. She

still uses this "clearing" tactic today. Any signs of panic, disorganized swallowing, or retching are indicators to step back to a previous texture, taste, method, or stage. I tried to keep aware of her preferences and readiness to move forward and adjusted accordingly. I let her dip a spoon, toy, or finger into things to taste—like pureed fruits, veggies, yogurt, or ice cream. We then worked up to stickier foods like cream cheese. Food texture was changed very gradually.

I tried to limit mealtimes to 30 minutes, although I went as long as 45 minutes when needed. Snack times would be 15 minutes or so. At first, she was completely disinterested in eating and even had some aversion, therefore it was quite challenging.

I made some very silly puppets only for mealtimes that "loved to eat." The puppets were her favorite part and proved pivotal to my program's success. They could get her to eat foods and quantities that no one else could! She was encouraged to eat as much as she wanted at each session. I tried to only change one thing at a time, like texture, utensil, or type of food, so as to not overwhelm.

Distractions were effective in keeping her cooperative and in her chair for longer periods as she progressed. A 30-minute episode of a cute television show, for example, allowed me to get more bites of food in without her being bored, fidgety, or resistant.

### After Mealtime:

I supplemented her oral calories with high calorie formula and other blenderized foods through her g-tube. I wanted her GI system to be accustomed to a variety of foods and liquids. I found it important to keep slowly stretching the stomach to promote its capacity without



### Continued:

## How One Mom Transitioned Her Child from G-Tube to Oral Feeds

overdoing it, of course, which could cause reflux. A direct subtraction of calories from tube-feedings at each oral feeding was done. I kept in mind: limit calories gradually while keeping fluids high and the stomach stretched to ensure hunger by the next meal. This was achieved by gradually diluting tube feedings with water.

#### In Summary:

Your child should be at a good, solid weight before starting a program like this, since there is likely to be some initial weight loss. My daughter slimmed-down, but I closely monitored her weight-to-height ratio and dismissed irrelevant standardized growth charts. As long as she was tracking along her personal growth curve, there was no call for concern.

I patiently applied a combination of techniques with an optimistic attitude to achieve our goal. Although, if I had to choose one thing that was paramount, it was the

puppets! I used them every day to keep eating fun, even after the transition was complete. She still recounts those puppets as one of her fondest childhood memories. The next most important thing was allowing her choices, such as what food to eat, how to eat it, which dish to use, and what food the puppet should "eat" next.

Use of her oral muscles every day to eat promoted strengthening in those muscles, and her oral coordination only got better with time. I had to remind myself that it was all going to be worth the effort in order to keep my confidence and perseverance strong. It was a commitment, that's for certain, and well worth it for us. By the time she was 2 years old, she was consuming all her nutrition by mouth and showing good growth. We had the g-tube removed not long after confirming it a success. It's been many years and she continues to thrive. She is extremely happy that we provided her the option of eating by mouth, definitely enjoys eating, and continues to maintain a good weight years later.



coordinate the muscles needed for safe eating and drinking by mouth.

See the Appendix to learn more about transitioning from tube to oral feeding, including a sample weaning treatment plan. Also, see the personal account on previous page of how one child with oral weakness and swallowing dysfunction eventually transitioned successfully from g-tube feedings to oral feedings.

### Ensure Safety When Eating

People with CM generally have difficulty controlling thin liquids, swallowing thicker textures, and chewing harder foods. There are several strategies that may work for your child under the guidance of a feeding specialist. Common suggestions include:

- Position yourself to make it easier to eat, such as your head being up higher from your stomach so that contents travel down with the help of gravity
- Ask your speech/feeding therapist or occupational therapist about modified utensils and other feeding aids for infants, toddlers, children, and/or adults, depending on your needs
- Use safe swallowing methods and avoid distracting activity, talking, or laughing while eating
- Make food easier to eat such as thinning if too dense, thickening runny liquids, peeling skins off, chopping food into very small pieces, and cooking food to its most tender state
- Thin liquids can be harder to control in the mouth and during swallowing, so your specialist may recommend a special cup or other means of making liquids easier to swallow.
- Avoid foods likely to be choked on that are hard, sticky, rubbery, or chunky (i.e. corn, nuts, mixed textures)
- Avoid mixing textures and consistencies (e.g. milk and crunchy cereal is more likely to cause aspiration)

Some people are still able to enjoy food by mouth even though they have a g-tube or PEG tube. Tube feedings sometimes serve to supplement calories and fluids taken mostly by mouth. Please ask your feeding specialist or speech therapist whether it is safe to eat/drink some amount by mouth.



- Allow plenty of time for meals; it will take someone with CM much longer to eat
- Taking more than 30-40 minutes for a meal may indicate more help is needed
- If eating meals is time consuming, allow more opportunities to eat over the day—for example six smaller meals a day may be tolerated better than three big meals, and can sometimes be a way to help increase calorie intake
- Make sure food is well-chewed before swallowing
- It might be helpful to alternate drinking in between bites of food
- Consider using other methods of eating (i.e. temporary feeding tube) if your child is weaker due to chronic fatigue or illness
- Select bottles with nipples/teats that a baby can latch onto easily (despite weak mouth muscles) and with a flow of milk appropriate for your baby's needs
- Avoid overfilling the mouth as large amounts are harder to control
- If your child is tired, stop the feeding
- Make sure the mouth is clear of food at the end of the meal
- Do not lie down or engage in too much activity for 30 minutes following a meal

### Suctioning

During feedings, people salivate more and someone with CM may drool more than usual. Suctioning the mouth with a portable suction device can help limit drooling, prevent the excessive saliva from entering the lungs, and remove any food or drink lingering in the mouth after a swallow.

## Sample Menu Ideas for Promoting Oral Feeding in Someone with CM

Developed by several care givers for children or adults with developmental readiness.

### PUREED DIET – No chewing required.

#### Breakfast

- Baby rice or oatmeal instant puree cereal constituted with whole milk, rice milk, or soy milk.
- Porridge, Farina, Cream of Wheat (more dense than baby cereals).
- Milk or milk substitute fortified with milk powder and/or whey protein to drink.

#### Snack

- Smooth yogurt (Greek is highest in protein).
- Smooth pudding made with whole milk or evaporated milk.

#### Lunch

- Smooth, creamy vegetable medley soup (add cornstarch for more calories and complex carbohydrates).
- Strained soup with olive oil and/or cornstarch (no chunks).
- Pureed legumes.
- Vegetable/fruit juice medley to drink (purchase or use high-quality juicer to make your own).

#### Snack

- Pureed low-acid fruit (add coconut milk for more calories).
- Commercial baby food fruit purees (add Greek yogurt for protein and calories).
- Fruit smoothie with ice cream or Greek yogurt (no fibrous fruits or pulp, like oranges or pineapple).
- Smooth ice cream, frozen custard, or milkshake.

#### Dinner

- Pureed fish (no bones) with olive oil.
- Pureed baby food sweet potatoes (add buttermilk or butter for more calories).
- Apricot, pear, or peach nectar/juice (low acid) to drink.

#### Snack

- Creamy cereal (same as fed for breakfast) with cornstarch mixed in (helps lessen overnight “fasting” concerns).
- Nutritional supplement drink to boost daily calories and nutrients, when needed.

### SOFT TEXTURED DIET – Minimal chewing involved for more advanced feeders.

#### Breakfast

- Oatmeal (add ground flax seed for calories and extra fiber).
- Scrambled eggs.
- Quiche or soufflé.
- Whole milk or milk substitute fortified with milk powder and whey protein powder to drink.

#### Snack

- Small curd cottage cheese.
- Cooked, soft, mashed, or diced avocado (no peel remnants or seeds).
- Diced cantaloupe or honeydew fruit (no peel remnants or seeds).

#### Lunch

- Finely chopped pasta (or Orzo) with butter or smooth sauce (no stringy cheese).
- Finely chopped, soft casserole (no tough meat or large chunks).
- Well-cooked, diced vegetables such

as carrots, beets, cauliflower (no stringy parts, stalks, or tough outer hull like corn).

- Fruit nectar/juice to drink.

#### Snack

- Mashed banana.
- Diced watermelon (no peel/seeds).
- Tapioca or rice pudding.

#### Dinner/Supper

- Very finely chopped/minced, moist, tender red meat or poultry (no fat, gristle, bones or cartilage chunks).
- Mashed potatoes thinned and moistened with milk, sour cream, butter, or smooth gravy.
- Mashed squash with butter or ground nuts for added fat.
- Vegetable/fruit juice medley to drink (purchase or make).

#### Snack

- Creamy cereal with cornstarch mixed in (helps lessen overnight “fasting” concerns).
- Granola soaked in whole milk or soy milk to completely soften.
- Commercial formula to boost calorie and nutrient intake as needed.

## Gastrointestinal Motility and GERD

Constipation, gastroesophageal reflux disease (GERD), and frequent vomiting are common GI issues in CM. There are treatments available that are usually effective.

### Chronic Constipation

Chronic constipation and difficulty having bowel movements in CM can lead to a poor appetite because you have a full feeling in your abdomen. This will eventually lead to unintentional weight loss. Additionally, intestines that are full can make it more difficult for the diaphragm to move during inhalation, which may lead to smaller lung volumes and breathing issues.

Slow stomach emptying and constipation are common in CM, mostly because of less overall movement that the person can do. Symptoms include:

- Nausea
- Vomiting
- Abdominal pain
- Cramping
- Bloating
- Diminished appetite
- Fatigue

Treatment includes:

- Increasing fluid intake
- Increasing fiber in your diet
- Positioning yourself upright during and for a half hour after meals
- Standing, if able (use a stander if appropriate)
- Moving and exercising regularly to the extent possible
- Using stool softeners or laxatives for constipation
- Using prokinetic medication for slow stomach emptying

Some people with CM depend on daily doses of laxatives to keep stools soft and allow for daily bowel movements. Ask your GI specialist about a maintenance dose that is appropriate for your chronic constipation.

You may need to allow extra time using the toilet. Some find that having something else to do while sitting on the toilet helps keep them there long enough for a bowel movement to occur. Some ideas include reading a book, singing, listening to music, or playing a game while seated on the toilet, particularly

for children.

Sometimes a person that is constipated will have liquid stool pass around the hardened stool and they appear to have a problem with diarrhea. Using your hand, you may be able to feel through the lower abdomen and identify a hard area within the person's body. Pushing on it may cause discomfort to the constipated person.

A stomach X-ray is the best way to know for sure if there is hardened stool blocking the intestines. The treatment for this is increasing your fluid intake and using a higher dose of laxatives, stool softeners, or enemas to remove the obstructing stool. Sometimes an intestinal clean-out, done at home, may be needed to remove hardened stool. The process is simple and usually provides needed relief quickly. Ask your GI specialist what products and process you should use at home to accomplish this.

Your bowel health is influenced by what you eat and drink. If you diverge from your normal diet for some reason, you will likely see changes in your bowel movements. Ideally, you should be able to empty your bowel each day or every other day. If you find you have not been able to have a bowel movement for three or more days, and laxatives are not helping, you should see your doctor. Constipation can lead to serious health problems if it goes on too long.

### Reflux (also called GERD)

Reflux is the movement of stomach contents up into the throat or nose. It causes a burning sensation for some people, but others may just feel something in their throats. It can inflame the throat lining over time and lead to other oral health problems.

Symptoms of reflux include (one or more may be present):

- Pain or burning in the upper belly, chest, or throat after eating/drinking
- Burping, spitting up, or vomiting during or after eating/drinking
- Bad breath odor from the stomach contents
- Sour taste in the mouth
- Feeling something in the back of the throat, particularly after eating or drinking
- Irritated throat or swollen nodes

- Mild airway constriction
- Aspiration leading to frequent lung infections

Reflux is treated using medications to decrease the amount of stomach acid and by keeping the person in a suitable position (usually upright) during meals and after eating for at least a half hour. If those methods are not successful, your GI specialist may prescribe a medication that speeds up stomach emptying.

Sometimes frequent, recurrent vomiting is a problem in those with CM leading to dehydration, aspiration, pneumonia, and/or weight loss. Anti-nausea medications can be helpful when frequent vomiting occurs outside of a stomach illness. Sometimes, symptoms can be severe enough to require an operation called a Nissen fundoplication, in which part of the stomach is wrapped around the esophagus to reinforce the valve that normally keeps stomach contents from going up. Surgery is only considered if all other options have failed.

## Drooling

Drooling occurs because the weakness in face and swallowing muscles prevents people with CM from closing their mouths and spontaneously swallowing their saliva. Sometimes drooling is a sign of a severe swallowing problem.

Lip strengthening exercises such as blowing bubbles, puckering, saying “oo,” and puffing your cheeks out with air may be helpful to some who are mildly to moderately affected. If there is too much space between the upper and lower jaw, then lip closure may not be possible unless that gap is corrected.

Medications such as scopolamine, tropicamide, and glycopyrrolate can be effective in reducing drooling, but they can also worsen constipation and thicken mucus in the lungs, making it harder to cough up. L-tyrosine has been shown to decrease drooling in some people with nemaline myopathy. As with all supplements there may be side effects, so please let your doctor know if you are taking L-tyrosine.

The use of Botox injections into the salivary glands for treatment of severe drooling has been shown to be effective as

a temporary solution with few side effects. It is important that your doctor understands that Botox should never be injected into the muscle of someone with CM when using Botox in the salivary glands to help with drooling. The injections should be made under ultrasound guidance.

Some things to watch for are thicker secretions as they dry up and possible changes in oral health. Repeated injections are often needed for maintenance. Good data on effectiveness and side effects in CM patients is still lacking, so discuss this with your specialist.

## Speech

The ability to speak clearly is often impacted by CM. Weak face muscles, low voice volume, misaligned jaw or teeth, narrow mouth, lack of velopharyngeal closure causing air to escape out the nose, slower or weakened tongue and lip movements, and difficulty controlling breath for speech are all contributors to the problem. An evaluation of speech and language ability should be done as early as possible, between one and three years of age.

A speech and language pathologist can not only help guide the development of speech abilities, but also recommend alternative ways to communicate. Having a way to communicate with caregivers and socialize is empowering to someone with CM that lacks the ability to speak clearly.

Communication also promotes development in many ways.

Some infants, toddlers, and children with CM are taught basic sign language, given they have use of their hands to make the signs for common words. As long as the family knows the same basic signs, they can communicate together.



Image credit - <http://www.livestrong.com/article/154898-the-use-of-bubbles-for-breathing-exercises/>

Oral motor therapy to strengthen and coordinate muscles, breath training, orthodontics, and special devices like a palatal lift appliance can help improve speech. To achieve the best outcome, the techniques, exercises, and equipment recommended to you should be used daily. It may take months or years, but many people with CM will have noticeable improvements in their speech using these methods



and some may have remarkable improvements.

Assistive communication devices (also called augmentative or alternative communication devices) provide another means of communication or can make it easier for someone with CM.

Weakness of the soft palate and throat muscles leads to a difference in the sound and understandability of one's speech caused by air leaking out the nose (hypernasality) during speech. When the soft tissue (soft palate) towards the back of your mouth doesn't contract and your throat muscles are too weak for contracting during certain speech sounds, this is called velopharyngeal incompetency (VPI). In people affected in this way, articulation is less precise and some speech sounds

may be impossible to produce, like /g/ and /k/ in the back of the throat or /b/ requiring a build-up of air pressure in the mouth.

A prosthodontist is a specialist that can design a device called a palatal lift appliance that fits like a retainer at the roof of your mouth to hold up your soft palate and improve your speech. It is best to begin working with a prosthodontist early (i.e. before entering school) if speech is possible and VPI present. In particular cases, an oral surgeon may decide to perform surgery to help improve speech, such as through a pharyngeal flap procedure.

A tracheostomy can also affect the ability to speak. If the trach

### How a Palatal Lift Appliance Works

—Written by Parent of a Child with CM

Hypernasal speech is the distortion of speech that occurs when too much air escapes through the nose when the soft palate is immobile or has very little movement. Hypernasal speech is a barrier to communication for many people with CM. One solution, a palatal lift, is described here.

First, an impression of the person's upper mouth is taken and used to create a mold for use by a specialty manufacturer to create a custom-fit retainer that has a "tail" that extends back under the soft palate to hold it up. Thin bands attached to the molars hold the retainer up in the correct position. The tail of the retainer holds up the soft palate to improve hypernasal speech. There is a balance to be achieved so that over-correction does not occur, which would make /m/ and /n/ sounds distorted (like when you have a stuffed-up nose). A good prosthodontist and speech pathologist can help with ensuring

the correction is satisfactory.

It is recommended to start early with its use, be consistent and well-disciplined, and have a qualified speech therapist involved. You can enlist a team (it may be termed the craniofacial team or cleft palate team) which is made up of a host of specialists (pediatric dentist, orthodontist, prosthodontist, oral surgeon, speech therapist, ENT, feeding specialists) to develop a treatment plan. Not all major hospital systems do this well, so ask around and get second or third opinions when needed.

The unresponsive soft palate creates a similar oral environment to that of a child with a cleft palate. Because cleft palates are relatively common in the general population, look for resources and specialists from this area of expertise, as they may be helpful to your child with CM that has hypernasal speech.

The retainer will feel weird, even intrusive, to the wearer at first. One suggestion is to put in a new retainer while lying down so it goes in more comfortably with the initial insertion. The usual recommendation is to wear it for gradually increasing periods of time each day, getting used to how it feels, as there is a sensation of something being in the throat. Once used to it, you might not feel it anymore and may be comfortable wearing it every day, all day. It is to be removed while sleeping, though. Some people are not able to wear the palatal lift during colds and other illnesses, due to airway passage blockage. As the child grows, new lift retainers will have to be made. Your health insurance might pay for one every two years, but check to find out.

has a balloon called a cuff that can be deflated for speaking, it will allow air to pass over the vocal cords. Speaking is harder with a trach, but some people with CM are able to do so with practice. A device called a speaking valve is a one-way valve used by some people with tracheostomies to speak more easily, but some have trouble getting enough air to breathe when the valve is in place. Some people with CM will never be able to speak without assistive devices; and some, not at all. However, a speech and language pathologist can help in finding other ways to communicate.

### Oral and Dental Care

The health of your teeth and mouth will have an effect on your overall health. Without good oral care, the bacteria in your mouth can reach high levels and cause infections.

The following is a summary of oral health complications and treatment:

#### Dental hygiene

- See a dentist twice a year starting at age 1.
- Brush teeth at least twice a day from time of first tooth, or after meals.
- Someone with low muscle tone may find it harder to clear the mouth of foodstuff. Regular brushing will also help prevent aspiration of any residual food left in the mouth after oral eating.
- Assist as needed and position the person for best access to the teeth.
- Use assistive devices to promote independent brushing.
- Use a mouth rinse if there are not swallow safety issues and you can do the swoosh and spit actions.
- If aspiration is a concern, use a soft foam oral mouth swab to clean around the mouth, gums, and tongue.

#### Hypersensitivity of mouth

- Pediatric dentists or occupational therapists can offer techniques for those who resist teeth brushing.

#### Misaligned jaw and teeth or crowded teeth

- An orthodontist may help with misaligned oral anatomy

(malocclusion). Take into account the amount of weakness and means of maintaining the correction long-term.

Sometimes in CM the mouth is quite narrow, so there is not enough space for the tongue or a full set of teeth to grow in straight. Orthodontic care beginning in early childhood can improve the limited space, prevent over-crowding problems, permit proper development of the jaw and teeth as a child grows, and improve self-esteem. Guided development of the jaw and teeth creates better alignment, which makes it easier to speak and safer to eat by mouth.

Receiving orthodontic care beginning in early childhood may provide enough correction without the need for risky

surgeries later. Sometimes retainers (either permanent or removable) are used indefinitely to maintain the correction achieved through orthodontia.

Meet with a specialized medical team experienced in treating oral and jaw/face (maxillofacial) problems in those with CM to find out what the best options are for your situation. This team may include a pediatric dentist, special needs orthodontist, prosthodontist,

oral surgeon accustomed to helping CM patients, speech therapist, ENT experienced with CM patients, and feeding specialist. In some hospital systems, they may be called cleft palate or craniofacial teams or specialists.

If your doctor or medical team recommends use of a general anesthetic to put you to sleep during a procedure, make sure they are aware of your CM and the possible risk of malignant hyperthermia. It is recommended that medical or dental procedures requiring anesthesia be done under the care of an experienced medical team in a hospital setting rather than an outpatient setting, so that any unexpected circumstances can be handled by a prepared medical team.

Orthodontia is not recommended for everyone with CM that has oral and jaw problems, however. In some people with severe CM and very weak facial muscles, the risks might outweigh the benefits, or the benefits may be short term if there is no way to maintain the correction.





### Benefits of Early Orthodontic Treatment in CM: What Our Experience Taught Us

—Written by Parent of a Child with CM

Orthodontics is the science and art of redesigning the genetically established growth pattern. Some patients/families may decide to wait until growth has completed to do any work, or may decide against any treatment. Our orthodontist believes that redirecting the growth pattern throughout childhood is ideal for those with CM and may be enough to avoid surgical reconstruction later.

In our experience with orthodontic treatment, I have to agree with our orthodontist. There are benefits to correcting jaw anomalies early that you might not think about, such as:

1. Teeth that do not line up cannot be used for chewing food.
2. A mouth that is too narrow will not allow the tongue to move about as it needs to during eating and speaking.
3. A mouth that is too narrow will cause crowding of teeth which leads to:
  - A. Self-esteem problems.
  - B. Difficulty cleaning the teeth.
  - C. Inefficient or unsafe chewing ability.
4. When the upper and lower jaws are misaligned, you might not achieve lip closure which leads to:
  - A. More drool falling out.
  - B. Difficulty articulating words so that others can understand you.
  - C. Self-esteem problems.

My child began seeing the craniofacial team at age four. She had a large open bite and her lower jaw jutted out too far. She also had a very narrow hard palate and immobile soft palate. The extended treatment plan included:

- An expander was placed to widen her hard palate.
- She was fitted with a well-designed palatal lift to help with her immobile soft palate and improve her speech intelligibility (prosthodontics specialty).
- A bite block retainer was worn to help correct the open bite, later switched to molar cement, and finally to simple rubber bands to leverage her jaw and achieve proper alignment.
- Head gear was worn at night during childhood to pull the upper jaw in line with her lower jaw very slowly over time.
- Regular braces on her teeth as a teenager to guide them into position.

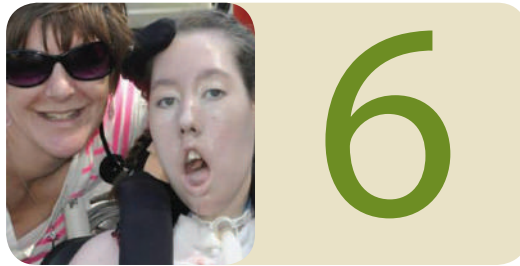
We can see that without the orthodontic treatment, her craniofacial proportions would be so far off by now that eating, having a conversation, or feeling good about herself would be severely corrupted.

For us, it has been the right decision to begin orthodontic treatment early and stick with it during her growth and development. To maintain the correction, she has a bracket behind her lower teeth that is not noticeable. A removable, upper retainer is worn as needed to maintain the correction of her upper teeth.



### Appendix Items

- Optimizing Body Weight
- Principles of Transitioning from Tube to Oral Feedings
- Types of Feeding Tubes



# Understanding Genetics & Testing

Our chromosomes exist in every cell of our bodies. Each dark band on a chromosome is a cluster of genes. Stated another way, chromosomes carry the genes. Our genes are made up of DNA. DNA has its own special alphabet made of four letters: A, T, G, C. There are at least 2,000 of those letters (called bases) to make just one gene. Some genes are made from hundreds of thousands of the letters. The order of these letters is the code for how proteins in the body will be made. Proteins control most every process, including how we grow and develop, what eye color we have, and how our muscles work.

One way to confirm the diagnosis of CM is to perform genetic testing, often done using a blood sample from the patient. The two most common genetic tests are sequencing and deletion/duplication testing. Sequencing reads through all the letters in the gene, looking for any errors or “misspellings.” Deletion/duplication testing looks for large chunks of code that are missing or extra.

There are many types of gene changes that can be identified through genetic testing. To better understand these changes, think of chromosomes as chapters in a book, then genes would be sentences in the chapter. DNA is like the letters that make up the words in the chapter. Gene changes or mutations would be errors in the words.

There are four main types of gene changes:

1. Missense Mutation—This occurs when a letter or word is changed. Example: THE SKY IS BLUE > THE SKY IS BBUE or THE SKY IS CAR
2. Nonsense Mutation—This occurs when the instructions stop too soon. Example: THE SKY IS BLUE > THE SKY
3. Insertion—This occurs when a letter or word is added.

Example: THE SKY IS BLUE > THE SKKY IS BLUE or THE SKY SKY IS BLUE

4. Deletion—This occurs when a letter or word is missing.

Example: THE SKY IS BLUE > THE SY IS BLUE or THE IS BLUE

Gene changes affect the way a protein is made. In CM, the protein that is affected is involved in helping the muscles work properly. If the protein is not made correctly, this will result in symptoms such as muscle weakness.

## Inheritance Patterns

Gene changes can be inherited from a family member(s) or may be spontaneous, meaning it is a new mutation that did not exist in either parent. Below are the common inheritance patterns and recurrence risks (risks to future pregnancies).

You can find some genetic terms defined at the end of this chapter and in the Glossary.

## Autosomal Dominant Inheritance

Autosomal means that both males and female exhibit symptoms of CM. Dominant means that just one of a pair of genes being miscoded is sufficient to cause symptoms of CM.

Recurrence Risks

For the parents of a child with CM:

- If one of the parents has CM:
  - ◊ 50% chance each pregnancy will result in another child with CM.
  - ◊ 50% chance each pregnancy will result in a child without CM. This unaffected child does not carry the

gene change so he/she cannot pass it on to his/her future children.

- If neither parent has CM and does not carry the gene change:
  - ◊ The risk to each subsequent pregnancy is the same as the general population, however, germline mosaicism cannot be ruled out.

**For the individual with CM:**

- 50% chance each pregnancy will result in a child with CM.
- 50% chance each pregnancy will result in a child without CM. This unaffected child does not carry the gene change so he/she cannot pass it on to his/her future children.

**Autosomal Recessive Inheritance**

Autosomal means that both males and females exhibit symptoms of CM. Recessive means that two copies of the gene change are required to have CM. Those who only carry one copy of the gene change are called carriers and do not typically have any symptoms.

Recurrence Risks:

**For the parents of a child with CM (both parents are**

**carriers):**

- 25% chance each pregnancy will result in another child with CM.
- 50% chance each pregnancy will result in a child who is a carrier, just like them.
- 25% chance each pregnancy will result in a child who does not carry the gene change and does not have CM. This unaffected child does not carry the gene change so he/she cannot pass it on to his/her future children.

**For the individual with CM:**

The risk to future children depends on the carrier status of your partner.

- If your partner has CM (and has changes in the same gene):
  - ◊ 100% chance to have a child with CM.
- If your partner is a carrier for CM (and carries a change in the same gene):
  - ◊ 50% chance each pregnancy will result in a child with CM.
  - ◊ 50% chance each pregnancy will result in a child who is a carrier, just like your partner.
- If your partner does not have CM and does not carry a gene change:
  - ◊ 100% chance to have a child who is a carrier.

**Understanding Genetics**  
 Visit [www.genetics.edu.au/](http://www.genetics.edu.au/) and click on Fact Sheets to learn more about patterns of inheritance, including Autosomal Recessive and Autosomal Dominant.

**X-linked Inheritance**

Males have one X chromosome and one Y chromosome. Females have two X chromosomes. When a gene change occurs on the X chromosome, males, having only one X chromosome, typically develop symptoms; whereas females have two X chromosomes, so they usually do not develop symptoms and are called carriers. Sometimes female carriers do have symptoms of the disease, but this does not occur often.

Recurrence Risks:

**For the parents of a child with CM:**

- If the mother is a carrier of CM:
  - ◊ Sons:



- » 50% chance each pregnancy will result in a son with CM.
- » 50% chance each pregnancy will result in a son without CM.
- ◇ Daughters:
  - » 50% chance each pregnancy will result in a daughter who is a carrier, like her mother.
  - » 50% chance each pregnancy will result in a daughter who does not carry the gene change and does not have CM. She does not carry the gene change, so she cannot pass it on to her future children.
- If the father has CM:
  - ◇ 100% chance to have a daughter who is a carrier
  - ◇ 0% chance to have a son with CM. The father will pass on his Y chromosome to his son which does not have the gene change.
- If the mother is not a carrier and the father does not have CM:
  - ◇ The risk each pregnancy will result in CM is the same as the general population, however, germline mosaicism cannot be ruled out.

## Family Planning

Many people wonder what the future may hold for them and their family members, especially when deciding how many children to have. Getting your genetic mutation identified is the first step in family planning with CM. Once you and your doctor know the mutation, you can better estimate the level of risk each pregnancy may have based on the inheritance pattern. When the risk is substantial to a family, they can discuss pre-pregnancy options with a genetic counselor, such as in vitro fertilization (IVF) or adoption.

## Testing Options for Future Pregnancies

Once the inheritance pattern of CM and the gene change(s) have been identified, then testing for future pregnancies can be done. Below are the testing options.

### Prior to a Pregnancy

- Preimplantation Genetic Diagnosis—PGD uses in vitro fertilization (the sperm and egg are combined in the lab).



The embryos are tested for the gene change identified in the family member with CM. Only embryos without the gene change are implanted. This prevents the child from having CM.

### During a Pregnancy

- Chorionic Villus Sampling—CVS is typically performed at 10-12 weeks of pregnancy. A piece of placenta is taken for genetic testing to see if the baby carries the gene change identified in the family member with CM. Since this is an invasive procedure, it carries a risk for miscarriage.
- Amniocentesis—An amnio is typically performed at 15-20 weeks of pregnancy. Amniotic fluid, the fluid around the baby, is taken for genetic testing to see if the baby carries the gene change identified in the family member with CM. Since this is an invasive procedure, it carries a risk for miscarriage.
- Non-invasive Prenatal Testing—NIPT is typically performed at 10-22 weeks of pregnancy. A blood sample is taken from the mother. This technology allows the doctors to separate the babies' cells from the mother's cells circulating in her blood. The baby's cells can then be tested for the gene change identified in the family member with a genetic condition. As of 2014, NIPT is used to screen for chromosome abnormalities such as Down syndrome, but in the future it may be able to detect CM.

### Genetic Terms

These are common terms that may be used to explain why the inheritance pattern is not straightforward.

**Mosaicism:** a proportion of cells carry the gene change. Think of a mosaic tile wall with different colored tiles.

**Somatic Mosaicism:** a proportion of cells in the body carry the gene change. The gene change occurred after fertilization, so only some cells carry the gene change. This can explain why symptoms are different in different parts of the body.

**Germline Mosaicism:** a proportion of egg or sperm cells carry the gene change. This can explain why a mother or father of a child with CM is not a carrier, but they have multiple children with CM.

**Reduced Penetrance:** the gene change is not expressed in an individual, even though they carry the gene change identified in the family.

**Variable Expressivity:** different members in the family have different symptoms even though they all have the same gene change.

**Skewed X-inactivation:** women have two X chromosomes in each cell of the body. However, only one X needs to be active in each cell. The pattern of which X is active and which is inactive

is usually random. However, in some women, one of the X chromosomes is preferentially active. This can explain why women can develop symptoms of an X-linked condition.

**Manifesting Carrier:** symptomatic carriers are sometimes called a manifesting carrier.



### Appendix Items

- Research in Congenital Myopathy
- Congenital Myopathy by Gene-Related Subtype





# 7

## Transition to Adulthood

After being taken care of by a team of pediatric specialists who have become familiar with your care, switching to a group of adult specialists can cause worry and doubt. This is very understandable, but with some preparation, the transition can be smooth. Adult neuromuscular multidisciplinary teams function in a very similar manner as the pediatric teams. Most multidisciplinary teams consists of a cardiologist, neurologist, pulmonologist, wheelchair specialist and rehabilitation specialists that either work together in the same clinic or in collaboration, although at different clinic times. These specialists should also be experienced in neuromuscular disorders that are traditionally thought to be childhood disorders.

Before becoming an adult, it is a good idea to gradually take on more responsibility for your healthcare, such as refilling your own medications or scheduling your own appointments. Caregivers have been handling all of the responsibility for many years, so it is a big change for them, too. During medical appointments, caregivers could begin looking to the teenage child to answer the nurse's or doctor's questions during the initial stages of transitioning toward adult care.

Choosing your primary care provider is important. This person should be easy to reach and respond quickly to your questions and requests. This person will need to be your advocate.

It is important for you to actively participate in your health

care. Perhaps you were participating even before, but when you become a legal adult, you are directly responsible for the decisions made. Information sharing among the team will benefit everyone—and most importantly, you. However, your medical team will be restricted in what information they can share with your family about your healthcare once you are an adult.

If speech is difficult for you, it may be useful to have someone accompany you to medical appointments that is familiar with your unique speech pattern to help ensure the medical team understands you and your needs. Technology can also help, such as having a text to speech device with you that can read out loud the statements that you type. When the primary issue with others understanding your speech is hypernasality, you could try plugging your nose while you speak to direct the airflow out of your mouth to better produce speech sounds.

CM is not a progressive condition in which muscle is being destroyed, but you can experience changes in your condition over time. Do not assume that every new or changing symptom is related to CM, however. People with CM are just as susceptible to the same diseases and age-related declines that all people face.

Following are some topics you may want to consider.



## 1. Health Insurance

The following applies to countries without national health insurance.

### Considerations

- Your employer may offer health insurance
- You might be able to obtain health insurance through a government program
- Some might be able to get health insurance through a parent's provider
- If your healthcare costs are very high, you might consider purchasing your own health insurance

### Recommendations

- Meet with a social worker to discover your options
- If you are working, ask your supervisor or human resources specialist about health insurance through work



## 2. Driving

### Considerations

- Weakness, movement limitations, and slower reflexes can impact your ability to drive
- Handicap accessible parking pass

### Recommendations

- Meet with a vocational rehabilitation specialist to learn about programs and driving options
- See if you could drive with vehicle modifications
- Plan for what mobility equipment you will travel with when deciding upon a vehicle to purchase

## 3. Transportation

### Considerations

- Public transportation may be accessible to you and more feasible or convenient than driving
- Ride-share programs
- Volunteer drivers
- Medical transportation company
- Travel reimbursement for medical appointments

### Recommendations

- Meet with a social worker and/or vocational rehab specialist to learn about available options



### My Decision to Live on Campus During College

One of the most important decisions you'll make in your life is whether or not to go to college after high school,

and if you choose to go that route, which college to attend. For someone growing up with nemaline myopathy, however, an equally important decision is whether to go away to college, or stay at home. It's a decision that I struggled with when I was 17, as my parents had been my support system for my entire life, and I had no idea what to expect without them.

When I had finally decided on my college, a small private school about 30 minutes from my home, my parents encouraged me to live at college. They said, "You need to get that college experience." If I didn't like it for the first year, I could come back and live at home for the remaining years.

Given that everyone's disability affects them differently, please think of all the pros and cons to this decision before making it. But if it is at all feasible, I strongly advise you to live on campus at your college.

## 4. Community Life

### Considerations

- Goal setting
- Finding opportunities
- Volunteering
- Socializing
- Financial management
- High health care costs
- Safe living arrangements
- Level of independence achieved
- Home nursing care
- Assistance through government programs
- Self-advocacy
- University, training, or job
- Scholarship availability
- Letters of reference from teachers, doctors, therapists
- Skill-building for interviews
- Relationships and sexuality

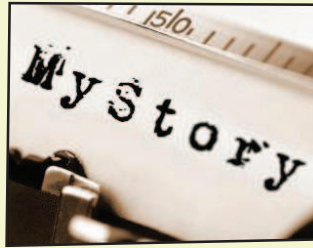
### Recommendations

- Pursue your dreams and goals
- Go to reduced-cost clinics and use preventive care to minimize costs
- Living with family, friends, or roommates will cut living costs and they could help you with daily needs
- Explore assisted living options
- Higher education and employment can enhance self-esteem while bringing monetary and social rewards
- Use vocational programs that help disabled clients get jobs within their physical abilities
- Speak with your doctor or counselor about relationship or sexuality matters

## 5. Adult Healthcare

### Considerations

- Weakness might increase due to having a larger, heavier adult body size and through aging
- It's possible to develop a new disorder independent of CM at any time
- Diabetes, obesity, heart attack, and stroke are more prevalent in people as they age



### Doctor: "It's Probably Just Your Muscle Disorder"

When someone has a muscle disorder, doctors may forget that they can also have common medical conditions, perhaps related to age or gender. Here are the thoughts of one adult living

with CM on this subject: "I hate that a lot of doctors blame everything on my muscle disorder. So, every time I had a sore toe it was because of my muscle disorder. Gained/lost weight, it was the muscle disorder. Had a pain in my side, it was my muscle disorder, but actually it was a tumor! When something goes wrong with you, they should treat you like everyone else and look for the real cause but not just throw up their hands and say, "Well, it must be your muscle disorder getting worse."

- Mental wellness
- Family planning
- Create an [Advance Directive for Medical Decisions](#) (AKA Wishes document)

### Recommendations

- Exercise and eat healthy to prevent obesity and minimize other health problems
- Get screened for potential health concerns to catch them early
- Take care of your emotional needs and seek counseling when you need help
- Obtain copies of your medical records
- Consider pursuing genetic confirmation of your CM if not done yet
- Seek genetic counseling if planning for a family
- Discuss birth control with your doctor before becoming sexually active
- As you plan for having children, discuss pregnancy-related risks with your doctor
- See an occupational therapist about developing child care skills if you are adopting or having children of your own
- An Advance Directive will help you explore with a doctor how you would like to be treated if you become unable to make decisions yourself

## Getting and Keeping Your First Job When You Have CM

### Basic skills starting at a young age:

- Begin self-advocacy development as early as possible in the child's life.
- Practice social skills with your child early and regularly.

### Skill building during high school:

- Practice talking about your disability so that you can explain it in the most positive light out in the community.
- Start with conversations about what would be her/his dream job and then make it realistic based on abilities and budding skill sets.
- If she/he wants to be a professional swimmer, but that is not feasible, ask what she likes about it and find skill sets that mesh with a job goal of professional swimmer.
- Use the IEP as a planning tool for her future plans, taking the right courses, getting experiences that will help pave the way, networking through school relationships to meet the right people.
- Join community groups, volunteer, and get to know people of influence. Who you know will get you more opportunities than anything else.
- Exploration period: find out what you are good at and where that overlaps with what you like to do.
- Family involvement and support in the process is critical, maintain high expectations for youths while keeping it realistic.
- Work on skills at home: phone skills, social situations, budgeting, organizing a shopping list, managing a household, etc.
- Volunteer at the place you want to have a paid job and you'll have your "foot in the door" while waiting for an internship opportunity or career.
- Job site accommodations can be simple: instead of lowering a water fountain, just install a cup dispenser next to it at a low height.
- If you will need job accommodations, state those up front at the interview.
- Sharing your diagnosis with an employer is only appropriate if you are obviously disabled or need accommodations. Employers are not allowed to ask about a disability.
- Practice what questions may come up about your disability and how to answer them in the most positive way.
- Employers often find that their employees with disabilities are the best because they appreciate what they have more than many other employees.
- Survey employers to find out what they are looking for in young people that are potential new hires, things like being reliable, punctual, honest, having good communication skills, willing to learn new things, being flexible.





## Coping with a Diagnosis of CM

Initial feelings with a new CM diagnosis can often include shock, emotional numbness, denial, anger, and fear for the future. Many will ask the rhetorical question: Why has this happened? It is normal to go through a period of uncertainty and emotional difficulty, but it is possible for families to find workable coping strategies.

Families, including extended members, differ in how they manage this new challenge. Some families rally to support, while others find it difficult to cope or may withdraw. This also poses special challenges within a marriage. Spouses may wrongly blame each other. Some couples may find it brings them closer. Grandparents may have trouble accepting the new family dynamics. Young siblings may feel scared or cheated. The primary caregiver will have overwhelming pressures. The affected person and the entire family will have many challenges ahead.

First and foremost, accept that you are going to have a different family life from what you had planned or expected. Hopes and dreams of the family can be redefined. Keep in mind that these expectations may change as a person's function changes over time. Trust in yourself to get through it. Seek support from people in a similar situation that will understand you. Learn how to become a caregiver that you never expected to be. Establish a routine. Keep learning. Find joy in the simplest of things.

Some good ways to control stress:

- Make time for yourself to relax or do something you love.
- Make time to connect with others that you care about (coffee, lunch date, support group, etc.).
- Accept help when it's offered.

- Ask for help when you need it.
- Talk about your day, stressors, feelings, etc.
- Have a stretching routine.
- Meditate.
- Take a walk or exercise.
- Sleep when you need it.
- Maintain a healthy diet.
- Stay organized, keep to a schedule, make lists.
- It is ok to seek professional counseling or medical support as needed.

Coping and acceptance of a CM diagnosis can change over time. Many find that the time period right after the initial diagnosis is the most challenging emotionally. Finding the coping strategies that work best takes time. A happy, joyful family life is possible for people with CM.

Often, the best support comes from developing relationships with other people who have been there. Even if they don't share the same diagnosis you're dealing with, many of the feelings they have experienced are the same. Online groups for your diagnosis, as well as local support groups for people with disabilities and families, can be invaluable resources.

The remaining portion of this chapter includes a collection of personal stories and interviews of parents and people living with congenital myopathies. They have shared their thoughts and feelings in hopes that you and your family can find some encouragement and ideas as you move forward in your own journey.



## On Mental Health and Wellness

A collection of statements from parents and people whose lives are affected by CM.

### 1. Facing Multiple Stressors

I feel that for new parents struggling with their child's myopathy and the many complications that arise, it is difficult to "stay afloat" emotionally.

First of all, it's physically exhausting to care for kids on a vent – the daily respiratory interventions take time as do the tube feeds. Even in the hospital, parents can't "relax" because they can't always trust the care. The myopathy is a grieving process – some people can handle this independently but others benefit from more help. I'm glad there are coping groups that parents can attend if they choose to do so. Mental wellness means being able to enjoy your daily life no matter what; being able to cope effectively with multiple stressors.

### 2. Finding Peace Through Talking Openly

Being able to talk about the challenges at hand openly to someone helps me a great deal. I notice a great relief when I have voiced whatever is heavy on my heart or mind. It's like peace has settled in – not expecting someone will have all the answers but an open ear means the world to me. It's what has gotten me this far.

### 3. An Honest Account of the Struggles

I hate going out in public on my own and I avoid it at all costs, I suffer with anxiety and panic attacks in public due to the sheer volume of strangers who

find my disability so hilarious they just can't help staring or making fun. I have depression, as well. I rarely think about my disability until others point it out. Knowing soon I won't be able to pick up my own son and struggling to do things with him is very



upsetting. Not working is infuriating and now I worry about receiving health care coverage. Life is tough but I still love my life! I like being "different," but sometimes it really grinds me down.

### 4. The Pressures a Mom Feels

Sometimes I feel like I am going to drown under the stress I feel. But I suck it up and put on a happy face for

my family. I worry about my child ALL the time. Is she healthy enough, is she happy, what more can I do to make her life easier, am I a good enough mum, am I doing a good job home schooling, am I paying enough attention to my other responsibilities, etc, etc... People look at my daughter and say "Wow, she looks so good!" But no one knows the blood, sweat, and tears that have gone into getting her here. I am afraid for the future, but I am so scared to talk about it because I feel like if I start crying, I won't be able to stop. I wish more than anything that she is happy, no matter what life has in store for her.

### 5. Honoring a Child's Life

If you ever wonder what you can do to help grieving parents – remember their child. Don't worry about bringing up the subject, because, believe me, we think of our lost ones 24/7.



## How a Mother Finds Joy Despite Son's CM

**A blog entry from a mother who says writing is a therapeutic coping tool for her.**

As my son turns 2 today, I am thinking about the gift he has proven to be. He's been expressing his wants and needs more, but he doesn't ask for much. He loves our nighttime routine. After I hook up his continuous overnight feeding and put his clubfoot brace on, he signs "book" by placing his hands together and opening them up. Then he taps the spot beside him, making sure I will sit where I always do. As we read, he uses sign language to name the animals on each page. The lion has been his recent favorite. He runs his hands over his head and down for the mane and adds a growl for good measure. He just started to say a few words too. "Elmo" is his most complex spoken word, so we'll be celebrating today with an Elmo cake.

It's hard for him to raise his arms up above his head, but he reaches up for me when he hears music and wants to dance. Another part of today's celebration will include dancing in the living room. A true boy, he loves cars, trucks and trains. He also loves playing with his older sister, and even pulls her hair and throws things at her.

As I think back on the last two years, I picture a dark, uncertain and rocky landscape. But I also feel the joys that have surprised us along the way. Joys made so much sweeter due to the intensity, grief and fears that come when your child has a neuromuscular disorder. Now, we stop to recognize the fullness of everyday, typical moments that occur when raising a family. Moments I may have otherwise overlooked.

The joys have come in unexpected form. Like seeing my son sit on a stool and push himself to stand for the first time by holding onto the bathtub — just so he could play with his sister while she was taking

a bath. Or finally being able to feed him "real food" by blending baby food, yogurt, quinoa and chicken together and pushing it through his feeding tube. And then there's the parents we've met that we never otherwise would have, and being able to share a deep understanding.

Even the rocky places have sharpened and strengthened us. Like when my son was strapped down for a CatScan of his irregularly shaped head and he cried out, "Mama!" louder and clearer than I've ever heard. I cried the whole way home, but thankfully all was well. Or coming to a place where I'm finally content (most days) with letting what feels like a parade of nurses and therapists into our home. And being able to live with unresolved questions about what the next course of treatment will be for him. With prayer and careful research, it usually becomes clear.

Two years ago today, I had been through a night of difficult labor and still the baby was taking awhile. It was difficult from a mental standpoint more than a physical one, as I knew the baby inside me had clubbed feet, and I wondered if something more could be wrong. My husband was a continuous, patient support. I never could've imagined how our lives were about to be shaken. But I also couldn't have anticipated how my son's loving nature and perseverance through trials have helped us grab ahold of the sweetness of "this day," and how he pushes us to rejoice and be glad in it.



## Mom Makes Maintaining Family Life a Priority

**A short story from a mom of a high-tech special needs child with a serious muscle disease.**

My son was born in 2006 with very low muscle tone, a weak cry, and empty sacks of skin where muscles are supposed to be. The shock wave of such a birth, the expectation of a having a healthy baby completely dashed, put me into a fog of fright and fear that lasted for many days. No one knew what was wrong, but what they did know was that he had trouble breathing on his own, he could not close his mouth to suck, and he needed suction to help him keep saliva from going down his airway. Keeping vigil night and day, watching monitors in the NICU, was all I could do.

Coming home with him after two months was a grand occasion, full of excitement that we were making progress and anticipation that maybe some sense of normalcy might be on the horizon. What no one could prepare myself and my family for was the invasion of the small army of nurses, respiratory therapists, specialized equipment, physical therapists, and others. The buzzing of equipment that turned my son's room into a mini-intensive care unit. Nurses who would be in our home, at odd hours like 10PM, or during dinner. All this makes for a huge adjustment on the part of each family member, perhaps especially our other two children. They had less quality time and energy from me and their father, and the much-anticipated arrival of their baby sibling turned out to be worrisome and disappointing for them, I'm sure.

I was concerned about them feeling too much of the impact, so I made sure that each day I had a nurse come in the afternoon, so I could do things with them as I had before, like helping with homework or taking them to piano lessons. They held their brother and played with him now and then. Looking

back, I think I could have included them a little more in his daily care and routines, which might have helped them bond more. Nurses felt a bit obtrusive in our home, but at the same time, a necessity so that I could be freer. We were also very fortunate to have grandparents nearby who were happy to help with the two older ones, and I was only too glad to have them be in "Grandma's" loving care.

Our lives changed with regards to family vacations and outings. We made it work by having nurses come for Saturday afternoons so we could take the older children skiing in the winter, or out on the lake in the summertime. Trying to keep their lives as normal as possible was important to me. Bigger vacations and time away without our son was made possible by having a special nurse, with whom we had a very high degree of trust, take him to her house for a few days. We would pay her extra money from a respite fund, and we knew he would be safe and happy while we, either as a couple or as a family, got a much-needed and refreshing break.

Looking back after a number of years, I see that overall things went well for my family; but if there is anything that could have been done better, it would be to have seen a family counselor, as a family, as a couple, and as individuals. A counselor can help guide, focus, and even lead when needed. A counselor understands the difficulties you are encountering. Solving these challenges with some help instead of all on my own might have saved some hard struggles. Now with my son well into his teenage years, things have eased, but we still find challenges that need to be overcome.





## Woman with CM Takes Up Social Justice

An Interview with Lynda Roy, BA, MSW

Lynda Roy is a very busy person by any measure. She works at a community health centre that provides barrier-free access to people that live with spinal cord-related disabilities, including neuromuscular disorders. She also works for a violence prevention organization in a program for women with disabilities. Finally, she is teaching at George Brown College in Toronto in the Assaulted Women and Children's Advocacy Program. Phew! Catching up with this fast-paced social justice advocate to talk was an ambitious move and totally worth it. Oh, and she happens to be living with congenital myopathy.

### **1. What led you to this field as your occupation?**

It was a bit of a meandering path to my chosen field for a variety of reasons. What it came down to was that I have been most interested in social justice work and supporting communities to make positive social change. I found that the most innovative thinkers were community activists and academics living with disabilities. I discovered that I really enjoyed reading articles and books, and appreciating art that focuses on and celebrates disability, particularly as it pertains to identity, body image, and sexuality.

### **2. Mental wellness is an important issue, especially for those that have congenital myopathy. As someone living with CM, please share what mental wellness means with regard to various life stages.**

Growing up with CM, I think the hardest part for me was developing positive self-esteem. For me, CM was not only a physical disability but also a facial difference and the two are very distinct experiences. Society is often more forgiving of a physical disability and not so forgiving of a facial

difference, particularly with young women and girls. This negatively impacted my self-esteem and my body image for a very long time.

When you live with any type of physical difference, the focus growing up is often on what your body cannot do and not on celebrating the ways that young people learn to problem solve by doing things differently.

Independence is often narrowly defined, too. For example, often the messages we receive as children is that wheelchairs are not a way of being independent but a way of being dependent. This is not true for many of us with physical limitations.

As recently as two years ago, I decided to get my first power wheelchair. This was a huge decision for me and to this day I struggle with it despite the reality that in order to keep working I need a mobility device (not just a car) where I can cut down on my stress and fatigue and focus on my work. A power wheelchair has helped to create a different level of access for me in my community. I am still walking and still driving a car, but the wheelchair is another option that enhances my independence.

Also, I feel it's important to have people that have similar life experiences that you can connect with and discuss the issues that are impacting you. For many of us, living with CM means that we lose abilities and possibly even good health at an earlier age than our non-disabled peers. I think it is important to develop a network of people that have similar lived experiences as well as support from friends and family.



### **3. What is most damaging to mental wellness for someone with congenital myopathy?**

For anyone that lives with a physical disability, I think there are a lot of myths and stereotypes that still exist that can

create barriers to developing deep relationships. When you think about it, up until about 40 years ago, people with disabilities were still largely kept at home or institutionalized. This history meant that most non-disabled people never came into contact on a regular basis with people with disabilities to form deeper relationships. So, to this day, and in my own work, I see people living in isolation.

Poverty also plays a factor in this. Access isn't just about automatic doors and ramps; it's about financial barriers, too. The majority of people with disabilities are still underemployed or unemployed, and they often live well below the poverty level. What does this mean? It means self-selecting out of social events like dinner parties because of fear you'll have to reciprocate. It means not being able to get a gym membership or take a course. These are all things that foster meeting new people and developing new friendships.

### **4. How can people with myopathy improve their mental health at each life stage?**

Parents can play an important role in ensuring that their child has positive self-esteem. Talk openly and honestly about your child's disability. Listen carefully to what your child is asking and answer exactly what your child is asking. If they are asking, they are ready for the answer.

We all have heard negative things about what it is like to live with a disability, but that shouldn't dictate your perception of what it's like. In order to support children with disabilities it is important to "unlearn" these negative stereotypes and to appreciate and celebrate your child for all of her/

his uniqueness. Disability will always be a part of who your child is; but as long as it's not the defining characteristic of your child and other parts of your child's identity are celebrated, this is just fine.

For those of us who are employed, I think a work/life balance is very important. I know I haven't always been great at finding that balance for myself, but I know that I need to pay attention more to my body and to my moods. We have less reserve at the end of the day than people who do not live with CM.

I think a big part of mental well-being is keeping active. For many of us, having to make the daily grind to work can prove impossible for a variety of reasons. Volunteering is a really great way to pursue your passions, keep busy, and meet new like-minded people.

For me, I think what has contributed most to mental well-being is having diverse interests and good solid relationships with people around me. I enjoy having downtime with friends and close family members. I like to be around people that make me laugh. I don't want someone who is always talking to me about disability because that indicates to me that they are seeing me in a very one-dimensional way. At the same time, I do not want someone who pretends my disability doesn't exist. My experiences have been informed by many aspects of my identity, and disability is one of those pieces. Ignoring someone's disability feels odd and is somehow invalidating of an essential part of who we are, I think.

### **5. What else is important to understand about mental wellness in the context of congenital myopathy?**

I think that the social determinants of health and our level of access to housing, employment, and social and recreational activities plays a vital role in our mental well-being. This is no different for people living with congenital myopathies.

# Appendix



## CoughAssist Protocol, Adult

Please refer to the CoughAssist Product User Manual for complete product description, including indications and contraindications for use.

Once it has been determined that the CoughAssist treatment is clinically appropriate, the following can be used as a suggested protocol.

**Indications:** For use with any patient unable to cough or clear secretions effectively due to reduced peak expiratory flow.

Those who might benefit from the use of the CoughAssist MI-E include any patient with an ineffective cough due to muscular dystrophy, myasthenia gravis, poliomyelitis, or other neurologic disorder with some paralysis of the respiratory muscles, such as spinal cord injury. It may also be used to treat ineffective cough due to other bronchopulmonary diseases, such as emphysema, cystic fibrosis, and bronchiectasis. It is effective for both tracheated and non-invasively ventilated patients.

### **Contraindications:**

- Any patient with a history of bullous emphysema
- Susceptibility to pneumothorax or pneumomediastinum
- Recent barotraumas

The above contraindications should be carefully considered before use.

**Warnings and cautions:** Refer to the CoughAssist User Guide.

**Implementation of CoughAssist:** Attach CoughAssist user circuit to the CoughAssist output including a bacterial/viral filter, smoothbore tubing and an appropriate interface: mask, mouthpiece, or trach adapter. If a mask is used, it should be of appropriate size to provide for a tight seal.

Begin with inspiratory pressures between +10 and +15 cm H<sub>2</sub>O and expiratory pressures of between -10 and -15 cm H<sub>2</sub>O to allow an introduction/acclimation period to the device. Verify initial pressures or any changes in pressure requirements by occluding the circuit in a clean manner and cycling from inhale to exhale. This should be done several times while viewing the pressure gauge.

**Settings and modes:** Start with either the Manual Mode or Auto Mode.

For Manual Mode: slide the manual toggle from inhale to exhale 4-6 times holding the inhale pressure for 2-3 seconds, enough time to deliver a full deep breath, then rapidly to exhale for 2-3 seconds.

For Auto Mode: slide to the Auto Mode. Set the inhale time to 2-3 seconds and the exhale time to 2-3 seconds.

Slowly adjust the pressures upward with subsequent treatments by 5 cm H<sub>2</sub>O each cycle of 4-6 breaths until optimal pressures are reached to clear secretions. Typical inhale pressures may vary between 40 cm H<sub>2</sub>O to 70 cm H<sub>2</sub>O. Optimal inhale pressure may vary from patient to patient depending on lung and chest wall compliance. Optimal exhale pressure may vary between 40 cm H<sub>2</sub>O and 70 cm H<sub>2</sub>O.

**Treatment length and process:** A standard treatment consists of applying 4-6 consecutive cough cycles of insufflation/exsufflation.

Visible secretions should be removed via suction from mouth, tracheostomy tube, or tubing. User should then rest for 20-60 seconds and return to normal mode of ventilation and prescribed oxygen flow, if needed. The cough cycles, alternating with rest periods, can be repeated 4-6 times for a full treatment.

**Using with a tracheostomy:** The CoughAssist treatment can be applied through a tracheostomy tube by using a 15 mm trach adaptor or by adapting to inline suction catheter that would allow for easy removal of secretions from the trach tube. Higher exhale pressures may be required to overcome the increased resistance of a tracheostomy or endotracheal tube. When treating with the CoughAssist through a trach tube, it is advisable to use a means for trapping any secretions that may potentially accumulate in the treatment circuit. Standard water traps, sputum traps, or extension tubing with corrugated inner walls can serve well for this purpose.

## Extubation Plan

You might find yourself in a situation where you need to be intubated while hospitalized. This means that due to breathing difficulty, an endotracheal tube is put in through the mouth or nose and into the windpipe. Once the breathing difficulty has improved, the tube can be removed. The procedure to remove the tube is called extubation. When having a planned surgery, this should be something you discuss with your surgeon or anesthesiologist in advance.

There may be situations where getting extubated becomes complicated for your medical team. A respiratory emergency letter template along with a detailed protocol for how to be extubated to noninvasive ventilation can be found on the following pages. Adapted from the work of Drs. Schroth and Bach, the protocol is entitled, "Recommendations for Post-Operative Care and Extubation of Children and Adults with Neuromuscular Disease." Another resource is the article listed below. All of these could be printed and shared with your doctors.

"Extubation of patients with neuromuscular weakness: a new management paradigm." By Bach JR1, Gonçalves MR, Hamdani I, Winck JC. *Chest*. 2010 May;137(5):1033-9. doi: 10.1378/chest.09-2144. Epub 2009 Dec 29.

The authors (Bach *et al*) have found this protocol to be successful in preventing tracheostomy and maintaining noninvasive ventilation (i.e., a ventilator or bilevel positive airway pressure device connected to a face or nasal mask or mouthpiece). Since protocols like this may be unfamiliar to many physicians, you may need to educate your team about this approach. This education ideally will take place prior to planned events like surgeries. Before agreeing to a tracheostomy, consider asking your clinicians to please read the medical literature on the topic by referring them to the article.

Many physicians assume that if a patient is too weak to breathe on their own or to cough effectively, then a tracheostomy surgery is needed before extubation is possible. However, this may not be necessary in many situations.

The extubation article (Bach *et al*) is summarized below:

Extubation can be attempted whether or not the patient still requires breathing support. They will just switch from using the

ventilator via the endotracheal tube to noninvasive ventilation.

Before attempting to extubate, the person should meet the following criteria:

- No fever
- No supplemental oxygen required to maintain SaO<sub>2</sub>  $\geq$  95%
- Clear chest x-ray
- Any medications that suppress breathing are discontinued
- There is a decreased need for suctioning

If these criteria are met, then consider extubation to noninvasive ventilation with no supplemental oxygen.

The key factors in successfully extubating to noninvasive ventilation include:

- Weaning off oxygen to room air
- Careful monitoring of both oxygen and carbon dioxide levels
- Using the Cough Assist whenever SaO<sub>2</sub> drops below 95%

A pulse oximeter and/or capnograph will help the team to know when you need more secretion management or breathing support. If you run into further breathing trouble, try to eliminate mask leaks, increase pressure support and/or ventilator rate, or try using a volume cycled ventilator. If these methods have all been tried but you continue to have trouble maintaining oxygen above 95%, then you may need to be re-intubated and try again at a later time using the same methods.

It may be necessary to use the noninvasive breathing support for more hours in the day until you regain more of your usual strength. When discharged from the hospital, you may resume your typical use of breathing support. For example, if you were previously using breathing support at night, you would go back to that.

Hi! My name is \_\_\_\_\_.

I have \_\_\_\_\_.

When I have a respiratory infection or pneumonia, my weak cough makes it difficult to keep mucus from building up in my lungs. I use a Cough Assist Device to help bring up the mucus.

My Cough Assist Settings:			
Inspiratory Pressure:	+		cm H2O
Expiratory Pressure:	-		cm H2O
Inspiratory time:			sec
Expiratory time:			sec
Pause:			sec

If my oxygen saturations are less than 95%, then I need more frequent Cough Assist and possibly BIPAP (IPAP 14-19 cm H2O; EPAP 0-4 cm H2O) or nasal mask non-invasive ventilation (assist controlled volume ventilation).

Using just oxygen to treat low oxygen saturations can mask elevating CO2 and cause CO2 retention and respiratory acidosis! As a general rule, oxygen should be avoided!

BIPAP and nasal mask non-invasive ventilation can prevent the need for intubation, tracheostomy, and reverse hypoxemia and respiratory acidosis. If you feel that intubation is needed, please pass on the attached extubation protocol to the ICU doctors and respiratory therapists.

My Specialists:	
Neurologist:	Contact #:
Pulmonologist:	Contact #:
Respiratory Therapist:	Contact #:
Cardiologist:	Contact #:

## Recommendations for Post-Operative Care and Extubation of Children and Adults with Neuromuscular Disease

### BEFORE EXTUBATION:

For INTUBATED patients, q 4 hours airway clearance:

1. Cough Assist: 5 sets of 5 breaths;

Inspiratory Pressure:	+30 or +35 or +40 for 1 – 2 seconds;
Expiratory pressure:	– 30 or – 35 or – 40 for 1 – 2 seconds;
Pause:	1 – 2 seconds

2. ETT suctioning followed by bagging.
3. Vest Therapy or Chest PT or Percussor for \_\_\_\_ minutes (with albuterol if prescribed).
4. Cough Assist: 5 sets of 5 breaths as above.
5. ETT suctioning followed by bagging.

\* Cough Assist can be used every 10 minutes followed by ETT suctioning and bagging \*

CONSIDER EXTUBATION when the patient is:

- Afebrile
- NOT requiring supplemental O<sub>2</sub>
- CXR is without atelectasis or infiltrates
- Off all respiratory depressants
- Minimal secretions

### AFTER EXTUBATION:

EXTUBATE to nasal ventilation and NO supplemental O<sub>2</sub>.

(eg. BIPAP of (14-20)/(3-6) using spontaneous timed mode; backup rate = spontaneous rate)

After EXTUBATION, minimum of q 4 hours airway clearance:

1. Cough Assist may be needed as often as every 10 minutes.
2. Cough Assist: 5 sets of 5 breaths then suction then re-expand w/ inspiratory breath.
3. Vest Therapy or Chest PT or Percussor for \_\_\_\_ minutes (with albuterol as prescribed).
4. Cough Assist: 5 sets of 5 breaths then suction then re-expand w/ inspiratory breath.

Wean from nasal BIPAP or ventilation during the day as tolerated; GOAL: only with sleep.

Use Cough Assist if O<sub>2</sub> sats drop to < 95% acutely.

Wean airway clearance regimen to 2-4 times a day.

Adapted from Univ of Wisconsin (Mary Schroth MD) 12/20/04 handout; Bach JR et al, Spinal muscular atrophy type 1: a noninvasive respiratory management approach. Chest 2000; 117:1100. Bach JR et al, Extubation of patients with neuromuscular weakness: a new management paradigm. CHEST 2010; 173(5): 1033.

(ver.12.1.14; LFW/SP/DS)

## The Pros and Cons of Having a Tracheostomy

Written in collaboration by an adult living with a tracheostomy and three parents caring for children with CM.

The decision to get a tracheostomy for you or your loved one requires careful consideration. Oftentimes, the decision is being made under tremendous stress, such as during an urgent hospitalization, when decisions are even more difficult to make. Below are suggestions that you may find helpful to know as you think about getting and living with a tracheostomy.

### Some Pros:

- A trach may be a medical necessity to sustain life.
- Fairly short recovery time from the surgery.
- Easier access to a stable airway.
- Easier to suction out mucus plugs and secretions during infections.
- No need for intubation through the mouth (endotracheal tube).
- When used with other medical devices such as suction machine, nebulizers, vest, and cough assist, a trach may be more effective than non-invasive ventilation in clearing the airway.
- No breathing mask is needed on the face.



### Some Cons:

- There is recovery time from the surgical procedure.
- You may require nursing care in the home because of the trach.
- Your school may require a Registered Nurse to accompany you all day during school.
- Mucus plugs may become stuck inside or outside the trach tube, blocking the flow of air.
- Although suctioning the trach is not painful, it is not a pleasant experience.
- Suctioning can cause irritation of the airway and more mucus production.
- Sometimes granulation tissue can form at the stoma.
- The stoma can become infected.
- Sometimes the trach can prevent speaking ability or may result in "garbled" speech.
- In some, having a trach can cause you to have trouble swallowing, resulting in aspiration of food, saliva or liquids (causing pneumonia).
- The tracheostomy tube requires special attention, such as:
  - ◊ regular bronchial suctioning
  - ◊ site/wound care
  - ◊ cannula changes
- You must carry a suction machine and a bag with extra catheters, trach tubes, and trach ties with you at all times.
- Can lead to an increase in secretions and higher risk of infection. There are certain germs that commonly form around trach tubes, like pseudomonas.
- Extra caution is required during swimming and bathing so that water does not accidentally enter the airway.
- Can result in tracheomalacia (floppy airway).



## How can I encourage movement to help development of the body and respiratory system?

### Learning to Move

What are the benefits of movement?

- Helps to maximize potential strength.
- Helps to stimulate circulation and lung expansion.
- Helps child explore and learn about his/her body and the environment.
- Helps child to interact with others.
- It's fun.

What role do the muscles play?

- Maintain a steady posture (lying, sitting, standing).
- Steady and brace one part of the body while another part moves.
- Bend or straighten a joint (bend an elbow or turn your head).
- Overcome gravity.
- Overcome friction.
- Work against opposition to movement, like tight clothes or tight joints.
- Stimulate bone growth through muscle pull on attachments to bone.

How can I help my child to move?

- Reduce the hindering effects of gravity, friction, and opposition to movement and offer physical support to help steady your child as they move. By developing strength, confidence, and pleasure in moving in this way your child may gradually require less help.
- Supported in side lying – reduces effect of gravity on arm movements.
- Hand resting on a toy car or similar – the wheels reduce the friction and make arm movement easier.
- Moving on a slippery surface (lino/laminate) and in slippery clothes reduces friction.
- Slippery finger paints – friction free.
- Sliding hand up a chalky board is easier than lifting arm up unsupported.
- Unrestricting clothing for easier movement.
- Water – in a bowl (hand play), a bath, spa, pool, lake (held by caregiver).
- Aquatic therapy (hydrotherapy) with a physical therapist.

- Swimming recreationally.
- Standing frame.
- Horse riding simulator or hippotherapy.
- Massage (stretching muscle, developing muscle tone).
- Tickling.
- Dancing.
- Squeaky toys from a pet shop are often easier to squeak than children's toys.
- Switches – pressure pads needing only light pressure to make toys work.
- Piano pads.
- Air hockey – particularly small executive toy size.
- Ten pin bowling – use a shoot, as your child can still direct the ball even if they can't hold it.
- Blowing and reaching for bubbles.
- Vibrating toys.
- Frog jumping bouncer.
- Wheeled toys: board scooter, tricycle, alternative bikes.
- Use an overhead sling to encourage independent play and increase joint range-of-motion (Bach 2004).
- Vibration therapy.
- Electrical stimulation.
- Anti-gravity treadmill (for those at least 1.422 meters or 4 feet 8 inches tall).
- Encouraging them to move to look at or reach for objects – popping bubbles for example.

### How Can I Improve Lung Capacity?

Swimming

Most specialists agree, swimming is about the best activity for your body and lungs. Armour et. al. concluded that swimming exercise promotes increased inspiration and expiration, developing greater chest volume, and an increased number of alveoli, or number of air sacs per volume of lung tissue. (Armour



J, Donnelly PM, Bye PT, 1993. The large lungs of elite swimmers: an increased alveolar number? Eur Respir J 1 Feb;6(2):237-47)

### Resistive Breathing Exercisers

1. Volume incentive spirometer
2. Deep breathing exerciser spirometer
3. Breathing exerciser
4. Volumetric exerciser
5. Breath builder

### Deep Breathing Technique

1. Inhale deeply as your chest expands and your ribs lift upward.
2. Hold the breath for five seconds.
3. Exhale slowly.
4. Push on abdominals to get the last of the air out of your lungs.
5. Repeat the series for 5 minutes each day (if you feel lightheaded, stop).

### Breath/Air Stacking – Glossopharyngeal Breathing (GPB)

Both inspiratory and, indirectly, expiratory muscle function can be assisted by “air stacking,” technically known as “glossopharyngeal breathing” (GPB). For individuals with weak inspiratory muscles and no ability to breathe on their own, GPB can provide normal lung ventilation and perfect safety throughout the day without using a ventilator or in the event of sudden ventilator failure day or night.

The technique involves the use of the glottis (throat) to add to an inspiratory effort by projecting (gulping) boluses of air into the lungs. The glottis closes with each “gulp.” One breath usually consists of 6 to 9 gulps of 40 to 200 mL each. During the training period the efficiency of GPB can be monitored by spirometrically measuring the milliliters of air per gulp, gulps per breath, and breaths per minute. A training manual and numerous videos are available.

GPB is rarely useful in the presence of an indwelling tracheostomy tube. It cannot be used when the tube is uncapped as it is during tracheostomy IPPV, and even when capped, the gulped air tends to leak around the outer walls of the tube and out the stoma as airway volumes and pressures

increase during the GPB air stacking process.

### Other Breathing Exercise Tips:

- Change body position frequently (at least every two hours).
- Encouraging them to sit up, if safe to do so (adaptive chair as needed).
- Crying and laughing can be good (they help to exercise the lungs). Leave them to cry a little longer if they are safe/comfortable.
- Steam in the bath may also help, but keep them in a warm environment when out of the bath.
- Encouraging singing, such as to nursery rhymes, and clapping. Even if this is difficult, it encourages movement and breathing.
  - Any blowing toys/instruments.

Movement is important to help the body and lungs develop and stay healthier. Encourage movement as much as tolerated, never to the point of exhaustion though. If you or your child is unwell, resting takes precedence over movement until you are on the road to recovery.



## Tips for Managing a Scoliosis Surgery

Written by the Parent of a Child with CM

Our child with congenital myopathy went through a spinal fusion surgery at age 12. Here are some tips we learned from it:

### Research Your Doctor

If you or a loved one are starting to have worsening curvature of the spine, you may want to consider consulting with several spine surgeons prior to the need for surgery. Get a feel for if the doctor you are seeing will be a good fit. I looked for a doctor that had experience with doing surgery on people with neuromuscular disease as well as with transitioning to non-invasive ventilation after the surgery. So, do you research when it comes to selecting a surgeon.

### Preparation

There are many factors to determining when it is time to perform surgery. Considerations include age, growth, overall health, surgery schedule availability, and the schedule of the whole family. We had several months lead time to prepare for the surgery. During that time we had a lot of preparation to do like:

Making sure each parent was in optimal health. This included addressing any health issues of our own so that we could be the best caregivers we could be. For me, that meant losing a few pounds and getting strong by going to the gym regularly. The exercise also helped me to deal with the stress.

Optimizing nutrition of my child. Nutrition plays a critical role in healing and recovery. We really worked to increase caloric intake for a few extra pounds and increase the quality of food and supplementation. Our surgeon often refers families to a nutritionist for this purpose prior to surgery. Some families may opt to consider placing a feeding tube for this purpose.

My son started listening to relaxation tapes at bedtime. This was to prepare him to be able to use them as a pain management or sleep strategy in the hospital. It worked great. The night before the surgery, he was sound asleep in less than nine minutes! I loaded these onto my phone for use in the hospital. I had my own versions that I used in the ICU to help

catch 20 minutes of relaxation, enough to refresh me for more hours of caregiving.

### Rally Your Support

We identified key family and friends to help us during different phases of the surgery. For example, we had a family member who would bring us fresh, healthy food every day to the hospital after work. We kept two coolers in rotation to provide us as caregivers with healthy food and snacks (our son was not eating by mouth after surgery). Identify what will help caregivers remain strong both physically and mentally.

We also used a tool from the MDA to help caregivers called, "myMuscleTeam." This free, practical online tool provides a powerful way to harness the strength of your "Muscle Team" of family and friends. Easy to use, the care coordination site offers several features that are valuable to you and the people who care about you. You can create private, secure myMuscleTeam Web pages, post journal entries to keep friends and family members updated, and enter items into a "care coordination calendar" that enables you and/or your primary caregiver to seek help in areas where assistance is needed, including transportation to medical appointments, meal preparation, household chores and more. Visit <http://mda.org/services/finding-support/mymuscle-team> to learn more.

Look into what kind of housing is available at or near the surgical hospital. Our acute care hospital had converted a wing into "hotel-like" rooms for caregivers. That way one of us could sleep while one of us stayed with our son. It was much better than commuting. Another option is to see if there is a Ronald McDonald house where you might be able to stay. This was key to having a place to take a break and rest.

### Knowing What to Expect

Pre-Surgical Consult—You can arrange a pre-surgical consult with your doctor or nurse practitioner to review important health factors, what to expect, and possible surgery risks. We did a one hour phone conference 30 days before surgery, but

each doctor may do it differently. One thing the doctor told us was that we should expect our child's face and body to be extremely swollen after surgery. That was extremely helpful to know because his face just did not look the same. Knowing this in advance helped to reduce anxiety about his appearance.

Tour—Our hospital offered tours prior to surgery for both child and parents so we would know what to expect. There were also videos on the hospital website designed for different age groups.

Talk with others who have been through it—I spoke with five other families whose child had been through a spine surgery. It was extremely helpful to get tips and suggestions from others.

Any hospitalizations can be stressful for families, especially with a major surgery such as scoliosis surgery. Some advance planning can be extremely helpful. Wishing you the best!



## Optimizing Body Weight

Weight is an important factor in everyone's health and this is especially true in people with CM. It is important to track height and weight for people with CM regularly.

One way to track growth in children is to use a growth curve, which compares height and weight to typical children of the same gender and age. A child with CM will not likely follow a typical growth pattern, but you can establish a baseline for your child and watch the trend over time to make sure he/she maintains a healthy growth curve. You can ask your primary doctor for a copy of a growth chart to track this over time.

Body mass index (BMI) is a common way to measure weight in comparison to height in adults. This is also something that can be tracked over time.

Optimal weight is key for people with CM. Being overweight can add extra stress and health problems on the body. Some medications, such as prednisone, may lead to weight gain in people with CM. It is common for people with CM to be underweight. The body needs some body fat stored in order to function optimally, although staying at the lower end of the optimal range is best, especially for someone with weak muscles.

Being too thin can also limit the body's ability to fight illness. If you are underweight, here are some ideas that may help:

- Add fat to your foods, such as butter, olive oil, cream, mayo, nuts.
- Puree fruits into a smoothie so that you can drink the calories of five fruit servings (which would never all fit into your stomach at one time otherwise!).
- Enjoy a full-fat ice cream or frozen custard shake fortified with whey protein powder.
- Choose the higher calorie options when making food choices to get more into each bite and drink your calories whenever possible.
- Supplement meals with high-calorie drinks, commercial formulas, and shakes.
- Try high-calorie nut-based flour in your cooking and baking (purchased from Internet companies or make your own).
- Add chopped or pureed nuts to your meals.
- Consume calories frequently throughout the day (eating

during the night is an option).

- Drink more fruit juice instead of just water.
- Add powdered milk to milk or milk-like drinks to add calories.

When all else fails, prescription drugs with appetite-stimulation side-effects are worth a try. Please talk with your dietician or nutritionist for more tips on how to optimize your body weight.



## Principles of Transitioning from Tube to Oral Feedings

### Introduction

For some people with CM, a feeding tube is a temporary need for getting nutrition. For others, tube feedings will be a lifeline tool to maintain nutrition. Some people use a combination of both oral and tube feeds throughout their life. Deciding what plan is best should be made in collaboration with your medical team.

It is important that preparation for transitioning to oral feedings starts when the feeding tube is first placed. You should have an oral motor stimulation program that you do daily with each meal time while tube feeding. This helps avoid oral aversion so you can resume eating by mouth with fewer setbacks once it is safe to do so. You or your child need to associate the mouth with hunger being satisfied. For example: you feel hungry and you eat, feeling and tasting the food in your mouth, and subsequently your hunger is satisfied. However, people who are only tube fed need help making this connection.

### Oral Stimulation Ideas During Tube Feeding

- Use a silky piece of fabric to stroke the cheek, moving toward the lips.
- Use a nubby brush or soft toothbrush to gently stroke the cheek, working toward the lips.
- Use a popsicle (cold, wet, sweet) to rub small amounts on the lips and encourage the person to lick it.
- Use vibrating toys designed for the mouth such as a vibrating teether.
- Consult with a speech or occupational therapist for more ideas.

### Oral Exercise Ideas (if age appropriate)

- Stick tongue out and swallow to strengthen the swallowing muscles.
- Push tongue into each side of the cheek and into the roof of the mouth and encourage to lick lips.
- Lip exercises such as tight closing/pursing of lips.
- Use a fat straw and a piece of paper with the aim of sucking the paper onto the end of the straw, then releasing it after a few seconds.

- Practice “dry” swallows.

### Knowing When to Transition

Transition to oral feeding often requires a team approach. For children, the caregiver will serve as the main player. Teams may include a nutritionist/dietician, feeding/speech therapist, behavior therapist (if behaviors are a barrier to eating), a nurse, and/or physician with experience in transitioning children with CM from tube to oral feeds.

First, check for readiness. The following questions are considered:

1. Is it safe to feed based on video swallow study results and observation of oral-motor skills?
2. Has the person shown appropriate growth or maintained bodyweight on tube feeds?
3. How many calories does the person need to eat daily to continue growing or maintain body weight?
4. Are you and other caregivers ready to transition? Do you have the time to devote to transitioning?
5. Has the medical team given clearance to start oral feeds?
6. Are you trained and aware of the signs and what to do if a choking incident occurs?

One of the initial steps in transitioning is to safely promote hunger. Often a child will not show hunger until they have reached an appropriate weight for height, so that is the top priority.

The tube feeding schedule needs to be normalized into meals and snacks. After the feeding schedule is normalized, calories are decreased in gradual increments and that volume replaced with water to meet fluid needs and keep the stomach distended with each meal.

Each bolus feeding is then increased by about 5 cc every three to four days, but see what is best tolerated in your situation. In this way there is greater distention of the stomach as the volume slowly increases over time. A slow stretch of the stomach will enable the person to tolerate larger boluses and will help to prevent the gagging or choking during the oral feedings to come. It is important to monitor body weight and height during this transition.

## Have Patience and Persistence

It takes time to change feeding behaviors. The longer the time a person goes without eating by mouth the longer it may take to transition to oral feeding. It is important to take small steps, making sure she or he is comfortable with the pace of the transition process. The person has the benefit of using the tube to meet nutritional requirements at this point, so try to relax, provide positive feedback, and keep your expectations realistic.

Since oral feeding may have been a source of discomfort (i.e. choking, aspiration, severe reflux, vomiting, etc.), an oral feeding aversion may have developed. The person may have learned that there is discomfort and perhaps even pain associated with oral feeding and is anxious and afraid to have anything in or around the mouth. This can be overcome, but some may need the help of a behavioral or feeding specialist during the early phase of transitioning.

Finding a network of other people that have transitioned to oral feeds may provide you with much needed support. There are online support groups you could join and you might want to check with your feeding team about a locally organized support group.

Only remove the tube after you demonstrate that she or he can eat and drink adequate amounts to continue growth. Make sure she or he does not lose excessive weight with illness or during the winter cold and virus season before removing the tube.

## Sample Weaning Program from Tube to Oral Feedings

- Discontinue all nighttime continuous drip feedings.
- Discontinue all continuous drip feedings by day.
- Develop age-appropriate mealtime schedule for daytime bolus tube feeds.
- Transition from predigested formula containing enzymes (such as Nutramigen, Progestimil, Vivenex) onto standard stock formula.
- Introduce oral stimulation and feeding small tastes, just prior to tube feeding.
- Start at the cheek; nothing too scary.
- Avoid the tongue, which is sensitive to touch and may initiate gagging.

- Practice nasal breathing and active closing of the lips if possible.
- Develop a system to reward progress in little steps. For example, with children, use a special “mealtime toy bag” as a distraction and other enticements. These toys are only played with during meal times and they must be engaging toys that the child loves. Use positive activities and reinforcement, lots of praise, and a socially engaging approach during meal times.
- Modify only one variable for oral feeding at a time, such as texture, temperature, or utensil used if you are easily overwhelmed at meal times.
- Monitor caloric intake and weight gain or loss.
- Duration of bolus tube feeding should approximate the length of an oral feeding.
- Liquid and food fed orally should be calorie dense (up to 30 to 40 calories per ounce).
- Once oral feeding is safely established in moderate amounts, a direct subtraction from tube fed calories may begin.
- Look out for signs of respiratory distress in between feeds, and stop the program if necessary.
- Criteria of no gagging, retching, or vomiting to move forward in the program.
- Successful weaning from tube feeds depends upon the child being a good candidate and your adherence to the transitioning techniques.
- Success is positive anticipation of food, maintaining/gaining weight appropriately, no aspiration, gagging, retching, or vomiting, and eating 100 percent of calories orally.



## Types of Feeding Tubes

The type of feeding tube used depends on how long a person will be tube fed, whether or not the stomach or intestines can tolerate the needed volume of food, and other anatomical/medical considerations.

### NG (Naso Gastric)

Runs from nose to stomach.

#### NG-Tube Pros

- Non-surgical.
- Parents can learn how to place an NG tube at home.
- Easily removed when need be.
- Good for short-term tube feeding.

#### NG-Tube Cons

- Need to be changed every 1-3 weeks, rotating sides of the nose.
- Easily pulled out.
- Uncomfortable.
- May contribute to swallowing problems and oral aversion.
- Lowers the cough reflex.
- Tube causes more secretions, which can be aspirated.
- Tube is very narrow and only very thin liquids and blends will go through.
- Tube can clog.
- Taping the tube to stay can be difficult and sometimes there is a skin reaction from the tape.
- Nasal or eye congestion may occur on the side where the NG-tube is located.
- Might make reflux worse because the tube holds the stomach open to the esophagus.
- Very visible and may draw unwanted attention in public.
- Only good for short-term use. (If your child is going to be tube fed for longer than three months, consider a g-tube.)
- The tube can be accidentally misplaced into the lungs instead of the stomach, which can be dangerous.

### G (Gastric)

A g-tube is surgically placed directly into the stomach through the skin. Some hospitals will place a PEG or Bard g-tube initially

to form the stoma (hole) for two to three months and then transition to a button g-tube.

Some doctors will tell you that you need to have a Nissen Fundoplication at the same time, but this is not true.

#### G-Tube Pros

- Considered a minor procedure placed under anesthesia or possibly without, depending upon your hospital.
- Comfortable for the user.
- Stays in place well; difficult to pull out.
- Parents can learn how to change the tube at home.
- Buttons can last for months (sometimes years depending on the type) between changing.
- Inconspicuous (stays hidden under clothing and low profile buttons do not stick out very far).
- Great range of feeding options (can feed thicker liquids and foods since the tubing is wider in diameter).
- Variety of methods to deliver the food, including bolus feeding with gravity and a syringe, syringe feeding with a plunger, gravity feeding with a bag and pole, and pump feeding.
- G-tubes may be vented to release gas from the stomach. Think of it as belly burping.

#### G-Tube Cons

- Surgical procedure required to place a g-tube and the site is sore for days to weeks during healing.
- Wound healing post surgery may be problematic.
- Although difficult to accidentally remove, this can occur.
- The balloon holding it in place can deflate or malfunction.
- G-tubes can clog (always flush feeds and medications with 7 to 10 mls of water).
- Infection or granulation is possible around the site (little blisters and redness around the stoma).
- If you do have a Nissen done, the amount of gas to vent increases.

Continuous tube feedings should be stopped for at least an hour prior to procedures during which a patient may have to lie flat, such as radiation. If tube feeds will be done at night it is advisable to elevate the head to prevent reflux.



## NJ (Naso Jejunal)

Runs from nose to intestines.

### NJ-Tube Pros

- Non-surgical.
- Can serve as a trial run before moving to a more permanent tube, such as the GJ.
- Can be used when there are stomach issues since it bypasses the stomach.

### NJ-Tube Cons

- The tube needs to be placed by Interventional Radiology using X-rays to insure correct placement into the intestines. Many hospitals perform this procedure under sedation, while some larger children's hospitals do not. You will need to check with your hospital.
- Your child will be continuously fed. There is no bolus feeding to the intestines. Your child can be off as many as six to eight hours. Feeding schedules vary based on nutrition and hydration needs.
- It is a bigger deal if your child pulls out the tube because you will need to get it placed by Interventional Radiology.
- In addition, there are all the same cons as for an NG.

## ND (Naso Duodenal)

Runs from nose to entrance of intestines.

### ND Pros and Cons

- Not a common tube and has similar pros and cons to the NJ-tube.

## GJ (Gastric Jejunal)

Button is placed at stomach and tubing runs to stomach (for venting and giving medicine) and also to small intestine for feeding. GJs come in short (children's) and long (adult) sizes. Longer lengths tend to stay in place better and can be used in children. Button GJs are much easier to work with in children.

### GJ-Tube Pros

- Feeds directly to the intestines (important for those with severe reflux, vomiting, delayed gastric emptying, dysmotility, or poor stomach function).
- A previously used g-tube can be converted into a GJ easily

(no new surgery).

- No more vomiting formula fed by tube (exception might be children with Chronic Intestinal Pseudo Obstruction and if tube has a complication).
- GJs can be an alternative to a Nissen for children who aspirate.

### GJ-Tube Cons

- Tube is placed by Interventional Radiology using X-rays and may be performed under anesthesia.
- GJs need to be replaced about every three months.
- Button should be kept stationary to avoid displacement (as opposed to the g-tube which should be rotated).
- GJs can coil up and migrate to the stomach if the J part isn't long enough. Seek medical attention immediately if you see formula in G output or venting.
- GJ-tube clogs are problematic if they occur. (Flush tube every four to six hours with 20 mLs of water through a syringe. This should be done less quickly than a flush to the g-tube, as water going through quickly can affect placement).
- Continuous pump feeds are required. There is no bolus feeding to a GJ because the small intestine cannot stretch like the stomach can. Feeding schedules vary based on nutrition and hydration needs, and if a child is able to eat or drink anything orally.
- Venting the G port is required.
- In rare cases, continuously venting or draining of the stomach is needed.

## J (Jejunal)

Button is surgically placed directly into the small intestine or into the stomach like a GJ is done.

### J-Tube Pros

See GJ-Tube Pros.

### J-Tube Cons

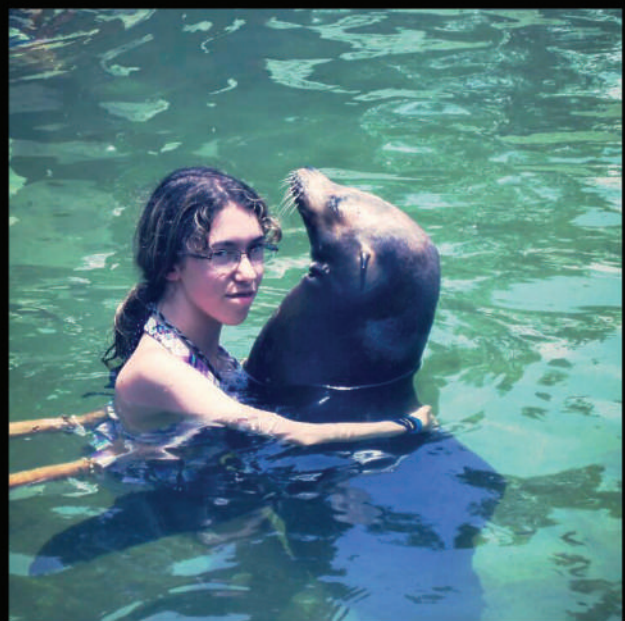
- J-tubes without G ports cannot be vented, so if there is build up of gas in the stomach it cannot be released.
- J-tube sites can have more problems with leakage than g-tube sites.

### TPN: Total Parenteral Nutrition

In rare cases when children are unable to tolerate tube feeding into their stomachs or intestines, or when they are unable to absorb nutrients from the GI tract sufficiently, TPN is used. TPN involves the placement of a central line and nutrition is fed intravenously directly into the bloodstream.

In some cases it is used temporarily until a child is able to tube feed again. In other cases, TPN becomes the main nutritional support. TPN can be administered at home once a parent or

caregiver is trained.



## Research in Congenital Myopathy

Congenital myopathy research is happening across the globe with collaborations among doctors, families, and scientists to understand, treat, and hopefully cure this group of diseases.

Since CM is rare, and some specific forms even rarer, participation of each and every eligible candidate is critical. Also, the more people who participate, the more meaningful the research can be. Smaller numbers of participants make it harder to prove if something works.

It can be challenging for researchers to enroll the number of participants required for a given study. Dropping out of a study, while sometimes unavoidable, leads to a change in population sample size which may affect the ability to provide answers to the study questions. All these factors make participation so crucial.

This section will briefly describe some different types of research trials and study categories:

**Natural History Studies**—this type of study gathers information on CM to better understand the natural course of the condition. Patients are evaluated at the beginning and then will be asked to follow-up to provide information on how the condition changes over time for the patient. The evaluation may include gathering medical records, having an exam done, doing functional assessments, etc. Natural history studies are useful in preparation for future clinical trials when a treatment is being tested. Without understanding the natural course of the condition first, it is difficult to know if the treatment being tested is the most likely contributor of an improvement in the condition.

**Tissue Repositories**—these are places to store tissues from patients that can be used in research studies and are sometimes called biobanks. The Congenital Muscle Disease Tissue Repository (CMD-TR) is one such place and aims to centralize tissue to safeguard it from loss, and distribute specimens to researchers conducting studies. Individuals and families can consent to have leftover muscle biopsy tissue, spare surgical tissue, or autopsy tissue sent to the CMD-TR. Contact Stacy Cossette at [scossette@mcw.edu](mailto:scossette@mcw.edu) for more information.

**Registries**—an important way to count and identify individuals

and families with a specific health condition. It includes gathering current contact information and in some cases, health information. The information is collected, stored, and used only in ways that respect people's privacy. Registries are a key part of a structure that supports researchers, doctors, and pharmaceutical companies to work towards a better understanding of CM and future treatments. The Congenital Muscle Disease International Registry (CMDIR) includes all congenital muscle disease subtypes. Visit [cmdir.org](http://cmdir.org) to learn more and register yourself or someone you care for with CM.

**Clinical Studies**—this type of study tries to answer a specific medical question about CM. For example, "Does CM cause heart failure?" This could be done by enrolling people and testing them to find the answer. The type of tests done will depend on the question being asked. Some studies do not require your direct participation and instead will look into your medical record to gather the necessary information.

**Clinical Trials**—this type of study involves an intervention. The intervention could be a medication, surgery, exercise plan, etc. In general, trials have a 'placebo' group and a 'treatment' group to see if the intervention is working. Placebo means that you are not getting the actual treatment and are instead receiving a sugar pill or a therapy not intended to do anything. Most trials will assign you randomly into either group. Also, neither you nor the trial doctor may know which group you belong to. Clinical trials are very helpful because they remove bias when testing an intervention and can tell us if it is truly effective.

**Basic Science Research**—this type of research is typically done in a lab and aims to understand the fundamentals of CM. This type of research typically uses animal models of CM to identify and test medications and other options for treatment.

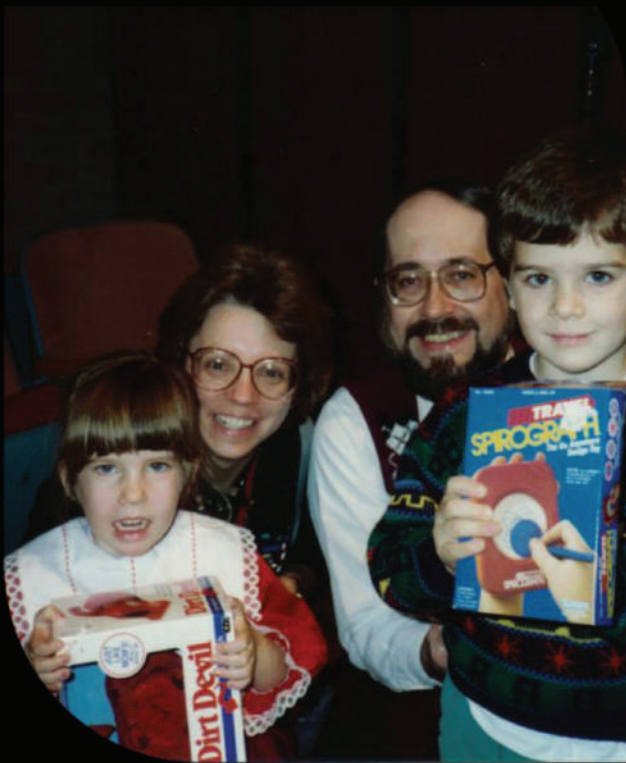
**Translational Research**—this type of research aims to apply the knowledge gained from basic science research onto patients with CM.

**Gene Therapy**—this is a type of translational research where genetic knowledge can be used to treat a disease. The goal of gene therapy is to provide patients with a healthy copy of the gene that is causing CM. The body can use the healthy gene to make a functional protein.

Enzyme replacement therapy—this is a type of translational research where knowledge of how proteins are made and function is used to treat CM. The goal is to provide a functional protein (ie. enzyme) for the body to use.



love is so nice



## CM by Gene-Related Subtype

This table shows a snapshot of what is documented based upon current findings at time of publication.

Gene symbol	Gene Subtype	Protein Deficiency	Muscle Biopsy Subtype Name
<i>ACTA1</i>	<i>ACTA1</i> -related myopathy	Alpha actin, skeletal muscle	Nemaline myopathy
			Cap myopathy*
			Congenital fiber type disproportion
<i>BIN1</i>	<i>BIN1</i> -related myopathy	Amphiphysin	Centronuclear myopathy
<i>CFL2</i>	<i>CFL2</i> -related myopathy	Cofilin 2 (muscle)	Nemaline myopathy
<i>CCDC78</i>	<i>CCDC78</i> -related myopathy	Coiled-Coil Domain-Containing Protein 78	Centronuclear myopathy
<i>CNTN1</i>	<i>CNTN1</i> -related myopathy	Contactin-1	Compton-North congenital myopathy
<i>DNM2</i>	<i>DNM2</i> -related myopathy	Dynamin 2	Centronuclear myopathy
<i>KBTD13</i>	<i>KBTD13</i> -related myopathy	Kelch repeat and BTB (POZ) domain containing 13	Nemaline myopathy
<i>KLHL40</i>	<i>KLHL40</i> -related myopathy	Kelch-like protein 40	Nemaline myopathy
<i>KLHL41</i>	<i>KLHL41</i> -related myopathy	Kelch-like protein 41	Nemaline myopathy
<i>LMOD3</i>	<i>LMOD3</i> -related myopathy	Leiomodin-3	Nemaline myopathy
<i>MEGF10</i>	<i>MEGF10</i> -related myopathy	Multiple EGF-like-domains 10	Dystrophic features and minicores
<i>MTM1</i>	<i>MTM1</i> -related myopathy	Myotubularin	Centronuclear myopathy/Myotubular myopathy
<i>MYBPC3</i>	<i>MYBPC3</i> -related myopathy	Cardiac myosin binding protein-C	Central cores with hypertrophic cardiomyopathy
<i>MYF6</i>	<i>MYF6</i> -related myopathy		Centronuclear myopathy
<i>MYH2</i>	<i>MYH2</i> -related myopathy	Myosin, heavy polypeptide 2, skeletal muscle	Inclusion body myopathy
<i>MYH7</i>	<i>MYH7</i> -related myopathy	Myosin, heavy polypeptide 7, cardiac muscle, beta	Laing distal myopathy
			Myosin storage (hyaline body) myopathy
			Multiminicore disease
<i>MYL2</i>	<i>MYL2</i> -related myopathy	Myosin, light chain 2	Congenital fiber type disproportion
<i>NEB</i>	<i>NEB</i> -related myopathy	Nebulin	Nemaline myopathy
			Cap myopathy*
<i>RYR1</i>	<i>RYR1</i> -related myopathy	Ryanodine receptor 1, skeletal muscle	Centronuclear myopathy
			Congenital fiber type disproportion
			Multiminicore myopathy
			Central core myopathy
			Core-rod disease or variant of nemaline rod myopathy
<i>SEPN1</i>	<i>SEPN1</i> -related myopathy	Selenoprotein N1	Congenital fiber type disproportion
			Multiminicore myopathy
<i>TNNT1</i>	<i>TNNT1</i> -related myopathy	Slow troponin T	Nemaline myopathy
<i>TPM2</i>	<i>TPM2</i> -related myopathy	Tropomyosin 2 (beta)	Nemaline myopathy
			Cap myopathy*
			Congenital fiber type disproportion
<i>TPM3</i>	<i>TPM3</i> -related myopathy	Tropomyosin 3	Nemaline myopathy
			Cap myopathy*
			Congenital fiber type disproportion
<i>TRIM32</i>	<i>TRIM32</i> -related myopathy	Tripartite motif-containing 32	Sarcotubular myopathy
<i>TTN</i>	<i>TTN</i> -related myopathy	Titin	Centronuclear myopathy
			Centronuclear myopathy with cores

\*Cap myopathy is considered by many to be a variant of nemaline myopathy.

# Resources for Families



LOVE



## A Short List of Some Clinical Support Resources

Muscular Dystrophy Association (MDA)  
[mda.org](http://mda.org)

Muscular Dystrophy Australia  
[mdaustralia.org.au](http://mdaustralia.org.au)

Muscular Dystrophy Canada  
[muscle.ca](http://muscle.ca)

Muscular Dystrophy Campaign  
[muscular-dystrophy.org](http://muscular-dystrophy.org)

Deutsche Gesellschaft für Muskelerkrankungen  
[dgm.org](http://dgm.org)

MD Canada Respiratory Guide  
[muscle.ca/living-with-muscular-dystrophy/respiratory-care](http://muscle.ca/living-with-muscular-dystrophy/respiratory-care)

Malignant Hyperthermia Association of the United States  
[mhaus.org](http://mhaus.org)

Shriner's Hospitals (see website for application)  
[shrinershospitalsforchildren.org](http://shrinershospitalsforchildren.org)

Center for Noninvasive Mechanical Ventilation, University Hospital, Newark, New Jersey  
[theuniversityhospital.com/ventilation/index.shtml](http://theuniversityhospital.com/ventilation/index.shtml)

FAQ on VEPTR for Scoliosis  
[chop.edu/service/thoracic-insufficiency-syndrome-center/veptr-faqs.html](http://chop.edu/service/thoracic-insufficiency-syndrome-center/veptr-faqs.html)



## Affected Individual and Family Support

VEPTR Kids—closed group on Facebook  
[facebook.com/groups/56052176312/](https://facebook.com/groups/56052176312/)

Support and advocacy group for parents of kids using mechanical ventilation  
[kids-with-vents.blogspot.com](https://kids-with-vents.blogspot.com)

## Central Core and Minicore Disease

[facebook.com/groups/243087794204/](https://facebook.com/groups/243087794204/)

## Centronuclear and Myotubular Myopathy

[mtm-cnm.com](https://mtm-cnm.com)

[centronuclear.org.uk](https://centronuclear.org.uk)

[facebook.com/groups/MtmCnmFamilySupportandDiscussion](https://facebook.com/groups/MtmCnmFamilySupportandDiscussion)

[facebook.com/groups/mtmcmfamilyconference](https://facebook.com/groups/mtmcmfamilyconference)

The Information Point for Centronuclear and Myotubular Myopathy  
[centronuclear.org.uk](https://centronuclear.org.uk)

Joshua Frase Foundation—Based in the USA  
[www.joshuafrase.org](https://www.joshuafrase.org)

JFF Facebook Page  
[facebook.com/pages/Joshua-Frase-Foundation-supporting-Myotubular-Myopathy/132151230150159?ref=hl](https://facebook.com/pages/Joshua-Frase-Foundation-supporting-Myotubular-Myopathy/132151230150159?ref=hl)

Welcome Packet for MTM Families  
[joshuafrase.org/uploads/MTM%20FAMILY%20GUIDE%20FINAL%20VERSION.pdf](https://joshuafrase.org/uploads/MTM%20FAMILY%20GUIDE%20FINAL%20VERSION.pdf)

MTM-CNM Family Conference  
[mtm-cnm.com](https://mtm-cnm.com)

Myotubular Trust Foundation—Based in the U.K., they support worldwide efforts with a focus on myotubular myopathy research in Europe.  
[myotubulartrust.com](https://myotubulartrust.com)

[RYR1.org](https://RYR1.org)

[will-cure.org](https://will-cure.org)

## Congenital Fiber Type Disproportion

[facebook.com/groups/228939138710](https://facebook.com/groups/228939138710)



## Nemaline Myopathy

Nemaline Myopathy Welcome Letter

[scribd.com/doc/242743410/WelcomeLetter-Update-100914](https://scribd.com/doc/242743410/WelcomeLetter-Update-100914)

A Foundation Building Strength—Founded in 2008 by parents of a child with NM dedicated to finding a treatment for NM.  
[buildingstrength.org](http://buildingstrength.org)

Nemaline Myopathy Facebook Page—Maintained by an NM parent who is also a clinical researcher in congenital myopathy.  
[facebook.com/NemalineMyopathy](https://facebook.com/NemalineMyopathy)

Nemaline Myopathy Support Groups—Several have been established, some in different languages.

[facebook.com/groups/715627125115427](https://facebook.com/groups/715627125115427)

[facebook.com/groups/miopatianemalinica](https://facebook.com/groups/miopatianemalinica)

Nemaline Myopathy Website—Established in 1999. [nemaline.org](http://nemaline.org)

A Family's Blog: [gretabaier.blogspot.com](http://gretabaier.blogspot.com)



## Tracheostomy Resources

[tracheostomy.com](http://tracheostomy.com)

Aaron Bissell, the inspiration; Cynthia Bissell, Founder; Ann Schrooten contributor (Attorney and mother of child with muscular dystrophy)

[globaltrach.org](http://globaltrach.org)

A multidisciplinary team of physicians, nurses, respiratory therapists, speech therapists, and patients working together to disseminate best practices and improve outcomes around tracheostomy care.

[trachkids.org/medical-conditions](http://trachkids.org/medical-conditions)

[hopkinsmedicine.org/tracheostomy/about](http://hopkinsmedicine.org/tracheostomy/about)

Johns Hopkins general info

[youtube.com/watch?v=tsLLbun1Xl0](https://www.youtube.com/watch?v=tsLLbun1Xl0)

How to change trach tube (teenager demonstrates)

[youtube.com/watch?v=d2wSiWuTxoc](https://www.youtube.com/watch?v=d2wSiWuTxoc)

Comprehensive trach care and cleaning

[youtube.com/watch?v=qsktXLbBtNI](https://www.youtube.com/watch?v=qsktXLbBtNI)

Trach cleaning/change infants

[kids-with-vents.blogspot.com](http://kids-with-vents.blogspot.com)

Support and advocacy group for parents of kids using mechanical ventilation



## Equipment and Adaptive Aids

### Wheelchairs, Scooters, Feeding Pumps, Etc.

#### Internet-based sales and exchanges

- New and used equipment
  - ◊ [Ebay.com](http://Ebay.com)
- New equipment
  - ◊ [Amazon.com](http://Amazon.com)
- Secondhand medical equipment
  - ◊ [affordablemedequipment.com](http://affordablemedequipment.com)
  - ◊ [infinitecdme.org](http://infinitecdme.org)
  - ◊ [ifmobility.mcservices.com](http://ifmobility.mcservices.com)
  - ◊ [chariotsofhope.org](http://chariotsofhope.org)
  - ◊ [lifenets.org/wheelchair](http://lifenets.org/wheelchair)
  - ◊ [ucpwfh.org/about](http://ucpwfh.org/about)
  - ◊ [craigslist.com](http://craigslist.com)
  - ◊ [freecycle.org](http://freecycle.org)
  - ◊ [oley.org/equipexchange.html](http://oley.org/equipexchange.html) for feeding supplies

#### Local Providers

- Loan closet
- Local durable equipment sales office
- Easter Seals program
- Shriner's Hospitals
- Social service or disability service centers
- Muscular dystrophy clinic
- Pharmacies

#### Cough Assist – Mechanical In/Exsufflator

- Phillips Respironics
  - ◊ [healthcare.philips.com/main/homehealth/respiratory\\_care/coughassist/default.wpd](http://healthcare.philips.com/main/homehealth/respiratory_care/coughassist/default.wpd)
- Patient Guide to the Cough Assist
  - ◊ [curecmd.org/wp-content/uploads/resources/cough-assist-guide.pdf](http://curecmd.org/wp-content/uploads/resources/cough-assist-guide.pdf)

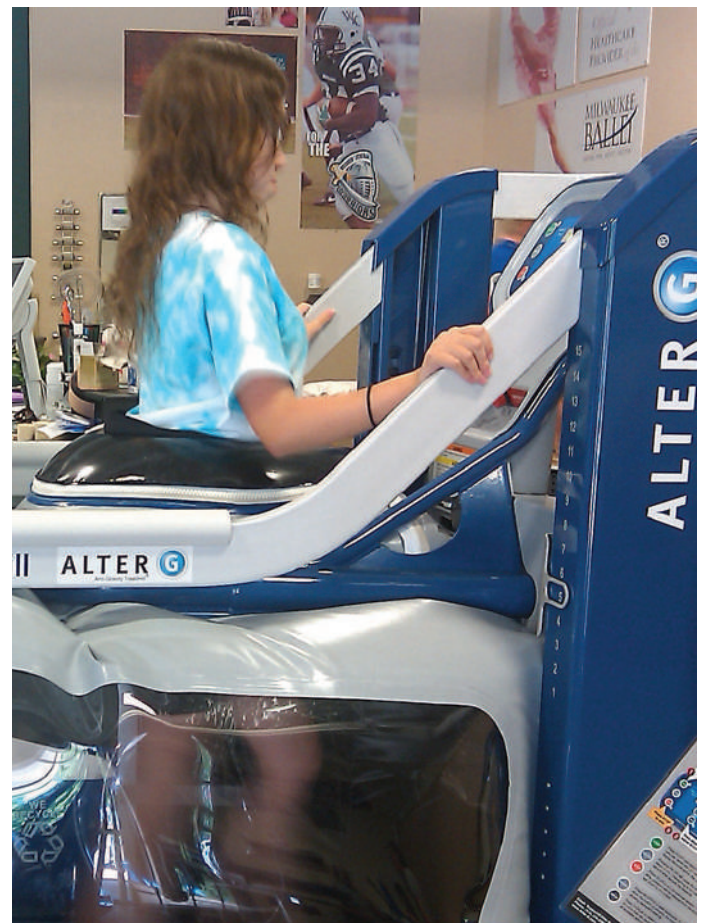
#### MedicAlert Foundation

- 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](http://www.medicalert.org)

## Non-Traditional Physical Therapy

Hippotherapy—Movement therapy on horseback.  
[americanhippotherapyassociation.org](http://americanhippotherapyassociation.org)

Alter G (Anti-Gravity) Treadmill—Safer way to gain or regain walking ability in later childhood or adulthood.  
[alterg.com](http://alterg.com)



## Tube Feeding Resources

[foodfortubies.com](http://foodfortubies.com)

Dedicated to sharing information and support for using blended food.

[facebook.com/foodfortubies](https://facebook.com/foodfortubies)

A not-for-profit group that shares information on blended diets for tube-fed persons.

[feedingtubeawareness.com](http://feedingtubeawareness.com)

General awareness and information sharing, especially for parents of tubie kids.

[mealtimeconnections.com](http://mealtimeconnections.com)

Brings together a group of professionals who have an understanding of complicated feeding concerns. Many resources and products available for purchase, including the *Homemade Blended Formula Handbook*.

[new-vis.com](http://new-vis.com)

New Visions provides continuing education and therapy services to professionals and parents working with infants and children with feeding, swallowing, oral-motor, and pre-speech problems.

[amazon.com/gp/product/1470190222](https://amazon.com/gp/product/1470190222)

*Complete Tubefeeding* is the definitive guide for anyone living with or preparing to receive a feeding tube, and those who care for them. It also includes a detailed discussion of blended diets.



# Glossary



**Abdominal** Relating to the abdomen, which is between your chest and pelvis.

**Achilles tendon** The tendon on the back of the ankle that attaches the heel to the leg.

**Acidosis** A condition in which there is too much acid in the body fluids.

**Active stretching** Where you assume a position and then hold it there with no assistance other than using the strength of your stretched muscles.

**Acute** An acute symptom is one that begins quickly or only lasts a short period of time.

**Advance directive for medical decisions** Advance directives are legal documents that allow you to explain your decisions about end-of-life care ahead of time. They give you a way to tell your wishes to family, friends, and health care professionals and to avoid confusion during a future time of failing health.

**Ambu<sup>®</sup> bag** A bag valve mask, abbreviated to BVM and sometimes known by the proprietary name Ambu bag or generically as a manual resuscitator or “self-inflating bag,” is a hand-held device commonly used to provide positive pressure ventilation to patients who are not breathing or not breathing adequately.

**Ambulatory** Relating to walking.

**Anesthesia** A drug or intervention to suppress pain to permit the performance of a surgery or procedure. Can be generalized or local.

**Apnea** The suspension or pause of external breathing. During apnea, there is no movement of the muscles of inhalation and the volume of the lungs initially remains unchanged.

**Arrhythmia** Any change or disturbance regarding heart beat.

**Arthrogyposis** The permanent fixation of a joint, present at birth, in a contracted position.

**Atrophy** The wasting away or degeneration of a body tissue or an organ.

**Augmentative or alternative communication devices (AAC)** Includes all forms of communication (other than oral

speech) that are used to express thoughts, needs, wants, and ideas.

**Autosomal dominant** Autosomal dominant means you need only one mutation/variant to have the disease. Autosomal dominant diseases usually have an affected parent, because the parent only carries one mutated copy of the gene which then gets passed to the child who has the disease. More often in families with a child with CM, the variant will be a *de novo* mutation, meaning it has occurred spontaneously in the child, but is not in either parent.

**Autosomal recessive** Both parents contribute a mutation/variant in the same gene to their child. The child must have both mutations to have CM. The parents are both carriers and are usually unaffected. The child may inherit a different mutation from each parent in the same gene (heterozygous). If the child inherits the same variant from both parents, it is called homozygous.

**Blood gas measurement** A blood test that is performed using blood from an artery to measure carbon dioxide content in the blood.

**Bolus feed** A single dose of a nutritional preparation given all at once.

**Botulinum toxin** A powerful neurotoxin that causes muscle paralysis and has been most widely used in cosmetic procedures to reduce wrinkles, but has been used to treat drooling problems in some people with CM.

**Breath stacking** A technique to expand lung capacity and to help form a productive cough for someone with weakened respiratory muscles. Can be performed with or without assistance, but basically involves taking a small breath, focusing on filling the base of your lungs, and holding for a second. Then, without exhaling, take in a larger breath and hold for one more second. Finally, take a big breath that fills your lungs as much as possible and hold for three to 10 seconds.

**Canine assistants** A dog specifically trained to help people who have disabilities including visual difficulties, hearing impairments, mental illnesses, seizures, diabetes, autism, or physical limitations.

**Capnograph** A device that monitors the concentration or partial pressure of carbon dioxide in respiratory gases.

**Cardiologist** A doctor who specializes in the study of the heart and its functions in health and disease.

**Cardiomyopathy** A group of chronic disorders affecting the muscle of the heart, resulting in impairment of the pumping function of the heart and can cause heart enlargement.

**Cells** The basic structural unit of all organisms. Usually microscopic in size, cells contain nuclear and cytoplasmic material enclosed by a semipermeable membrane.

**Central nuclei** Different from a normal muscle cell where nuclei are present along the outside of the cell, central nuclei are present within the muscle cell, directly in the center.

**Chronic** Something that is constant, lasting, or long-term.

**Clinical trial** A type of clinical research that follows a pre-defined plan or protocol. By taking part in clinical trials, participants can play a more active role in their own health care, access new treatments, and help others by contributing to medical research.

**CO<sub>2</sub>** Called carbon dioxide, this is the compound we exhale. It is a naturally-occurring chemical in the air given off by plants.

**Colon** The last part of the digestive system that extracts water and salt from solid wastes before they are eliminated from the body.

**Commercial formula** A combination of foods and liquids that have been thinned in a blender or food processor and strainer in a manufacturing plant. These foods and liquids can be eaten using a cup, straw, syringe, tube, or spoon.

**Congenital** Present at birth.

**Continuous feed** Nutritional formula that slowly drips through the feeding tube over several hours.

**Contractures** A permanent shortening of a muscle or joint that results in a decreased range of motion.

**Core** The trunk or torso. The central part of the body from which extend the neck and limbs.

**Cornea** The transparent front part of the eye that covers the iris, pupil, and anterior chamber.

**Dantrolene** A muscle relaxant used to treat malignant

hyperthermia that acts by abolishing excitation-contraction coupling in muscle cells, probably by action on the ryanodine receptor.

**De novo** De novo is a Latin expression meaning "from the beginning," and a de novo mutation is a genetic mutation that neither parent possessed nor transmitted.

**Deformity** A distortion or disfigurement of the body.

**Dehydrated** Severely lacking in water nourishment.

**Delirium** A syndrome that presents as severe confusion and disorientation, developing with relatively rapid onset and fluctuating in intensity.

**Diaphragm** The dome-shaped sheet of muscle and tendon that serves as the main muscle of respiration and plays a vital role in the breathing process.

**Dietician** A health care provider specializing in nutrition or dietetics.

**Discrepancy** A difference especially between things that should be the same.

**Dislocation** Putting out of joint or out of position, as a limb or organ.

**Disorder** An abnormal condition affecting the body.

**DNA** DNA stands for deoxyribonucleic acid. It is contained within the cell's nucleus in the form of chromosomes of nearly all living organisms. It is the carrier of genetic information from one generation to the next.

**Dual energy X-ray absorptiometry** Also known as DEXA. A means of measuring bone mineral density. Two X-ray beams with different energy levels are aimed at the patient's bones.

**Echocardiogram** An ultrasound of the heart. This test looks at the heart's structure and can help show how the heart is functioning.

**Electrocardiogram** A test that looks at the pattern and speed of the heartbeat. This test is performed by placing electrodes (monitors) on the chest, arms, and legs. Routine ECGs usually take less than one hour.

**Electrocardiographic** A device that detects and records the

minute differences in electric potential caused by heart action and occurring between different parts of the body.

**Electrolytes** Electrolytes are minerals in your blood and other body fluids that carry an electric charge. Electrolytes affect the amount of water in your body, the acidity of your blood (pH), your muscle function, and other important processes. You lose electrolytes when you sweat, throw up, or have watery stools. You must replace them by drinking fluids that contain electrolytes. Plain water does not contain electrolytes.

**Endotracheal tube** A specific type of tracheal tube that is usually inserted through the mouth or nose for emergency breathing assistance.

**Enema** A procedure in which liquid or gas is injected into the rectum, typically to expel its contents, but also to introduce drugs or permit X-ray imaging.

**ENT** A doctor who specializes in the ear, nose, and throat.

**Eustacian tube** A tube that links the nasopharynx to the middle ear.

**Failure to thrive** Indicates insufficient weight gain or inappropriate weight loss. This term is no longer in frequent use.

**Forced vital capacity (FVC)** Refers to 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.

**Gastroesophageal reflux disease (GERD)** A condition in which the stomach contents (food or liquid) leak backwards from the stomach into the esophagus (the tube from the mouth to the stomach). This action can irritate the esophagus, causing heartburn and other symptoms.

**Gastroenterologist** A doctor who specializes in the digestive system and its disorders.

**Gastrostomy or 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.

**Gene** A basic unit of heredity made up of DNA. Genes are

the blueprints or directions for how everything in your body is made. We inherit genes from our biological parents.

**Gene/Genetic mutation** A change in the DNA sequence of an organism's genome that can alter something in the body or how it functions.

**Genetic counselor** A health care provider who has a Master's degree with education and training in medical genetics and counseling. A genetic counselor can help explain which genetic mutation is causing your symptoms and may be able to help with predicting the likelihood of passing that mutation on to your children.

**Genetic testing** Genetic testing is among the newest and most sophisticated of techniques used to test for genetic disorders which involves direct examination of the DNA molecule itself.

**Geneticist** A doctor who studies genetics, the science of genes, heredity, and variation of organisms.

**Gross motor delay** The delay of crawling, sitting, walking, or other activities that use the large muscles.

**Growth curve** A curve on a graph in which weight and height are plotted to show growth for age over time.

**Heart failure** This occurs when the heart is unable to provide sufficient pump action to maintain blood flow to meet the needs of the body.

**Hoyer lift** A device with a motorized seat mechanism which enables it to lift the body from a sitting to a standing position or into other places.

**Hyperlaxity or Hypermobility** Joints that stretch farther than normal.

**Hypernasality** Another name for velopharyngeal insufficiency, a condition where the uvula doesn't properly close against the back of the throat, causing excess air to come out from the nose. It results in very nasally speech that can be difficult to understand.

**Hyporeflexia/areflexia** The condition of below normal or absent reflexes.

**Hypotonia** Low muscle tone.



**Inherited** A gene or characteristic that is passed down from parent to child.

**Intercostal** The muscles or intervals between the ribs.

**Interdisciplinary team** Health care providers from different specialties working together to see patients during a single consultation.

**Intestinal motility** The movement of waste spontaneously through the intestine.

**Intravenous (IV)** Administering a drug, nutrient solution, or other substance into the vein.

**Kyphosis** Atypical curvature of the upper back seen as a hump.

**Lethargy** A state of being drowsy, listless, unenergetic, or indifferent. Sluggish inactivity.

**Ligaments** The fibrous tissue that connects bones to other bones.

**Lordosis** Atypical forward curvature of the spine in the lumbar region, resulting in a swaybacked posture.

**Lung function** How well your lungs work.

**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 over-heat.

**Malignant hyperthermia treatment** Dantrolene is a muscle relaxant used to treat this.

**Malocclusion** Imperfect positioning of the teeth when the jaws are closed.

**Maxillofacial** Of or relating to the jaws and face.

**Mechanical ventilation** Assist with breathing using a ventilator.

**Microscopic appearance** Too small to be seen with the naked eye but large enough to be studied under a microscope.

**Milestones** Important skills expected at certain times during child development such as rolling over, sitting up, crawling, walking, talking, etc.

**Motor milestones** The physical skills expected to be achieved by children.

**Multidisciplinary team** A team approach that utilizes the skills and experience of individuals from different disciplines, with each discipline approaching the patient from their own perspective. Most often, this approach involves separate individual consultations.

**Muscle biopsy** A minor surgical procedure done under general or local anesthetic, using a needle or a small incision to remove a small sample of muscle. The procedure may be done to confirm a clinical diagnosis or rule out muscle disease.

**Myometer** An instrument for measuring the extent of a muscular contraction.

**Myopathy** Simply means muscle disease. A myopathy is a muscular disease in which the muscle fibers do not function properly resulting in muscular weakness.

**Nasogastric or NG-tube** A tube placed through the nasal passage to the stomach to provide nutrients or medication.

**Nasopharynx** The part of the pharynx behind and above the soft palate, directly continuous with the nasal passages.

**Negative inspiratory force** Negative Inspiratory Force (NIF) is the greatest force that the chest muscles can exert to take in a breath. The normal value is greater than -60. This number shows the doctor how strong your breathing muscles are.

**Nemaline rods** Abnormal accumulations of rod-shaped structures within muscle cells; when found, a diagnosis of nemaline myopathy is usually made.

**Neurologist and Neuromuscular doctor** A doctor who specializes in problems with the nervous system. The nervous system is broken down into the central nervous system (brain and spinal cord) and the peripheral nervous system (the connection between the nerves and the muscles). Neurologists treat different conditions, such as epilepsy, migraines, and developmental delays. Some neurologists have additional training and expertise in problems with the peripheral nervous system and the muscles (neuromuscular specialists).

**Neuromuscular disorder** A broad term that encompasses many diseases and ailments that impair the functioning of

the muscles.

**Nissen fundoplication** A surgical procedure to treat gastroesophageal reflux disease (GERD) and hiatal hernia.

**Nutritionist** A specialist who advises on matters of food and nutrition and the impacts on health.

**O<sub>2</sub>, O<sub>2</sub>, Oxygen** A naturally occurring chemical compound that all humans need to survive.

**Occupational therapist** A specialist with expertise in helping people make physical changes to their environments so that activities of daily living (such as eating, bathing, dressing, doing school work) are easier to do and persons can have greater independence.

**Ophthalmoplegia** Paralysis of the extracocular muscles that control the movements of the eye. Double vision is the symptom for this condition.

**Orthodontist** A doctor who specializes in straightening teeth and other jaw-related distortions.

**Orthopedist** A doctor who specializes in the prevention or correction of injuries or disorders of the skeletal system and associated muscles, joints, and ligaments.

**Orthotist** A specialist in the making and fitting of orthotic devices, such as braces to treat spinal curvature.

**Osteopenia** Reduced bone mass of lesser severity than osteoporosis.

**Osteoporosis** Medical condition in which the bones become brittle and fragile.

**Palatal lift appliance** A prosthetic device designed to fit against the hard palate, anchored by the teeth, with an extension along the soft palate to occlude part of the velopharyngeal opening. The device is used mainly by people with weak velopharyngeal musculature or excessively wide velopharyngeal openings to reduce nasal resonance and airflow during speech.

**Pectus Excavatum** An abnormal formation of the rib cage that gives the chest a caved-in or sunken appearance.

**Pharyngeal flap procedure** A surgical procedure to help correct or improve the quality of speech when hypernasality is

a problem.

**Physical therapist** A specialist who has a Master's (or higher) degree with expertise in helping people make physical changes to improve movement in the body. This includes proactive measures to help prevent the loss of movement through stretching or bracing.

**Placebo group** A group that is given a placebo in a research study. A placebo is something that does not directly affect the behavior or symptoms under study. A researcher must be able to separate placebo effects from the actual effects of the intervention being studied. For example, in a drug study, subjects in the experimental and placebo groups may receive identical-looking medication, but those in the experimental group are receiving the medicine while those in the placebo group are receiving a sugar pill.

**Pneumonia** An infection of the lungs that is caused by bacteria, viruses, fungi, or parasites. It is characterized primarily by inflammation of the alveoli in the lungs or by alveoli that are filled with fluid (alveoli are microscopic sacs in the lungs that absorb oxygen).

**Power of attorney for health care** A document that names your health care proxy. Your proxy is someone you trust to make health decisions for you if you are unable to do so.

**Primary diagnosis** The main diagnosis given to a patient.

**Prognosis** How the disorder is expected to change over time and what those changes mean for your child's health and life.

**Prosthodontist** A doctor that specializes in making dental prosthetics. Maxillofacial Prosthetics is a subspecialty of Prosthodontics that involves rehabilitation of patients with defects or disabilities that were present at birth or developed due to disease or trauma. Prostheses are often needed to replace missing areas of bone or tissue and restore oral functions such as swallowing, speech, and chewing.

**Protein** A class of organic compounds that consist of large molecules composed of long chains of amino acids. Proteins can be 1) functional like an enzyme that is responsible for almost every chemical process in the body; 2) structural, like hair, muscle, collagen, organs, etc.; or 3) antibodies.

**Psychologist** A doctor who specializes in diagnosing and treating problems related to thoughts, emotions, and behaviors.

**Ptosis** Ptosis is the drooping of the upper eyelid.

**Pulmonary** Relating to the lungs and respiratory system.

**Pulmonary Function Test (PFT)** 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.

**Pulmonologist** A doctor who specializes in problems of the lungs such as breathing issues or infection. Pulmonologists should work proactively with patients and their family to prevent complications from neuromuscular diseases.

**Pulse oximeter** A medical device that indirectly monitors the oxygen saturation of a patient's blood through a thin part of the patient's body, such as fingertip or earlobe.

**Pulse oximetry** A non-invasive method allowing the monitoring of the oxygen saturation of a patient's blood.

**Range of motion stretching** A term commonly used to refer to the movement of a joint from full flexion to full extension.

**Researcher** Someone who conducts research as in an organized and systematic investigation. Scientists are often described as researchers.

**Registry** Registries are a useful way to count and identify people with a specific health condition.

**Rehabilitation specialist or Physiatrist** A specialist to help individuals with physical and mental disabilities to integrate into society and live independently.

**Respiratory** Pertains to breathing.

**Scoliosis** An abnormal lateral or sideways curvature of the spine.

**Serial casting** A non-surgical approach aimed at reducing muscle tightness around a joint or resolving a contracture.

**Skeletal maturity** Determining the degree of maturation of a child's bones.

**Sleep study** May also be called Polysomnography.

Documenting what happens in a person's body during sleep. Many measurements are taken such as readings related to the lungs and heart. Brain function, along with eye movement and muscle movement, are monitored using different tests. End tidal CO<sub>2</sub> is important to measure during the study.

**Soft palate** The muscular part of the roof of the mouth, towards the back.

**Speaking valve** A plastic attachment that fits at the end of a cuffless tracheostomy tube or a tracheostomy tube with a deflated cuff. A speaking valve has a one-way valve that opens with inspiration and closes with expiration, causing the air to flow out past the vocal cords to facilitate speech.

**Speech therapist** A specialist who evaluates, diagnoses, treats, and helps to prevent disorders related to speech, language, communication, voice, swallowing, and fluency.

**Splinting** Application of a splint to keep an injured part rigid to avoid pain or further injury that may be caused by moving the part.

**Subluxation** When a bone comes partially out of a joint but does not completely dislocate.

**Subtypes** A subdivision of one type.

**Symptoms** A physical or mental feature that is regarded as indicating a condition of disease, particularly such a feature that is apparent to the patient.

**Tissue** A group of biological cells that perform a similar function.

**Torso** The trunk of the human body that excludes the head and limbs.

**Treatment group** A group of patients that is given a treatment/drug/intervention in a research study. A treatment is something that is hypothesized to directly affect the behavior or symptoms under study. For example, in a drug study, subjects in the treatment and placebo groups may receive identical-looking medication, but those in the treatment group are receiving the medicine while those in the placebo group are receiving a sugar pill.

**Velopharyngeal closure** Closure of the nasal airway by the elevation of the soft palate and contraction of the pharyngeal

walls. This closure is required for making vowel sounds and for all consonants except /n/, /m/, and /ng/.

**Velopharyngeal incompetency (VIP)** A disorder resulting in the improper closure of the velopharyngeal opening during speech, allowing air to escape through the nose instead of being directed through the mouth for correct speech sounds.

**Ventilator** A machine designed to mechanically move breatheable air into and out of the lungs to provide the mechanism of breathing for a patient who is physically unable to breathe or is breathing insufficiently.

**Video fluoroscopic swallow study** Also called a modified barium swallow exam (MBS) or swallow study, this is a radiologic examination of swallowing function that uses a special video X-ray called fluoroscopy.

**Vital capacity** The maximum amount of air a person can expel from the lungs after a maximum inhalation.



# The Care of Congenital Myopathy

## A Guide for Families

