

My Child Has CMT

A guide for parents of children
with Charcot-Marie-Tooth





- 1** The diagnosis
- 5** Questions and answers
- 15** One parent's story
- 19** Keeping a medical record
- 21** Medical terms explained
- 28** Where to get help

When a Child Is Diagnosed with CMT

Many parents report that the diagnosis of Charcot-Marie-Tooth disorder was just the beginning of a whole range of emotions they would eventually experience.

Most parents' emotions don't follow any set pattern or even last a predictable amount of time. They are, however, honest and valid responses to their family's circumstances. The father may be very accepting while the mother is very angry.

The important thing to remember is that all parents experience some emotions regarding their child's diagnosis and development. It is healthy, normal, and very common to feel angry, afraid, guilty, and sad. Once a parent can identify and acknowledge his/her feelings, he/she can begin to use some of the coping strategies that follow.

Denial and Disbelief

Some parents say that they had a feeling, long before the diagnosis was finally made, that something was wrong. In many cases, with a hereditary disorder, it was almost a situation of waiting for what they most feared: the same diagnosis that plagued their own youth. But, until the confirmation of their fears, they denied those feelings. This type of reaction is a very healthy emotion. It gives the person time to accept, adjust, and make plans. It can also protect a parent from too much pain, too soon.

Sadness and Depression

Many parents say that they experienced depression when their child was diagnosed with CMT. In some cases, if the child is the first family member to be diagnosed, the depression is a direct result of the ignorance of how serious the condition might become. Sadness can be tied into the dreams and expectations that the parents had for their child's life and their own.

Often, depression or depressed feelings may occur unexpectedly, without the person's even knowing why. Seeing other children doing something that your child cannot do or cannot do without pain and difficulty can trigger long-hidden feelings of sadness.



CMTA support
groups can help you
connect with other parents
whose children have the
same diagnosis.

Anger and Hostility

It is not uncommon for parents to direct their anger toward the doctor who diagnosed their child, or toward each other, the child, God, or close friends and family. Often, they will hear themselves saying, "It's not fair" or "Life isn't fair." It is perfectly normal to feel anger at the unjustness of having a disability.

Fear

Fear is a common reaction to the unknown. When parents are told that their child will not be “typical,” they feel apprehensive. When that general fear is coupled with the lack of a clear picture of what the future holds for someone with CMT, because of its variable nature, parents are often doubly fearful. It is normal to feel anxious about the uncertain future, to feel inadequate as a parent, to shield your child from pain and unhappiness, and to feel poorly prepared to meet your child’s needs, whatever they might be.

Learning to Cope

Undoubtedly, if you have a child with CMT, you have felt some or all of these emotions. Acknowledging your feelings and recognizing that other parents feel the same way will help you move toward accepting your child’s diagnosis and planning your child’s future. It is when a parent gets “stuck” in a particular emotional state, unable to move on, that feelings become counterproductive. Most parents have developed coping strategies to help them “move through” their different emotions and get on with other things.

Emotions are healthy responses to difficult situations. Using specific coping mechanisms will help you maintain a healthy balance in your life and allow you to deal with each situation in a productive and positive way.

Emotions do not magically “disappear” or go away at some point in time. Feelings occur throughout your life as you encounter new and stressful situations, make difficult transitions, or realize that your child has missed out on some milestone you had envisioned for him or her. At those times, it is important to acknowledge your feelings, allow yourself the permission to feel that way, and reach into a “bag” of coping strategies to help you deal with the situation. Then, you can, again, get on with your life.

WAYS TO COPE

Cognitive Coping

- Read everything you can on CMT
- Talk to the parent of a child with CMT
- Subscribe to newsletters on CMT or magazines like *Exceptional Parent*
- Attend workshops and conferences on CMT

Physical Coping

- Cry, laugh, have a sense of humor
- Exercise
- Eat well
- Rest sufficiently
- Take long walks
- Keep a normal routine

Psychological Coping

- Join a support group or start one
- Keep a journal
- Take up your child’s cause, by raising money or raising awareness
- Take one day at a time
- Replace negative thoughts with positive ones
- Mentally list all the wonderful things your child can do
- Realize you’re not alone
- Get counseling, if needed



Questions and Answers about CMT

By Mena Scavina, D.O., Neurologist and Co-Director, MDA Clinic, and Alisa Clark, R.N., M.S.N., PNP-BC, MDA Clinic Coordinator, Alfred I. DuPont Hospital for Children, Wilmington, Delaware

What will my child be able to do now?

It is important to remember that signs and symptoms vary greatly between individuals with CMT. This is often referred to as clinical heterogeneity, meaning that there are many variations on the theme! Even within a family in which the same gene is involved, there can be differences in strength, balance, upper or lower body involvement, etc. Having said that, children should be afforded the opportunity to try the activities they wish to participate in. After seeing how a child does with a particular activity, a decision can be made as to whether this is the right choice or not. Adaptations may need to be made as well. For example, playing goalie on the soccer team may be preferable to being the center forward so that the child does not have to run as much in that position and can still participate. Having a designated runner in baseball is another example that may allow a child to be part of a team with an acceptable accommodation. There are also organizations that specifically work with children who may need more adaptive activities. Examples are sled hockey or challenger baseball. Your community or healthcare provider may have more information regarding such groups.

What can we do to make things better—physical therapy, occupational therapy?

In our experience, physical and occupational therapy are key components in managing the symptoms and signs of CMT. Regular physical therapy for stretching and strengthening exercises as well as balance and gait training is very important. Occupational therapy is important for hand strengthening and fine motor skills. These services can be done privately as well as in the school system if educationally relevant. Proper shoes and appropriate orthotics can make all the difference in terms of safety and pain management. And as for all children and young adults, proper nutrition, weight management, adequate sleep habits, and regular physical activity are all part of good health.

How do you encourage kids to try most activities while drawing the line at activities that could prove harmful?

This is a very difficult question to answer and one that will vary based on parenting styles and beliefs. Most children do want to do things that their peers and siblings are doing. If possible, children should at least be given the opportunity to explore the activity at their own pace. If you or they feel they cannot safely take part in an activity, perhaps an alternative approach or adaptation can be found. However, children should be encouraged to find their strengths and weaknesses before they simply say no to an activity. There are many role models with physical limitations who achieve incredible physical acts. Children may benefit from seeing how others handle a specific physical limitation rather than simply giving up.

Is it normal for kids with CMT to complain of being tired all the time?

Fatigue is a real issue for individuals with underlying neuropathies and other neurological disorders. Children and adults frequently complain of fatigue with walking long distances such as at the mall or even at school going from class to class. However, should a sudden change in a child's level of activity occur, it is best to see your healthcare provider to make sure there are no other medical issues which may be contributing to the sudden change. Poor sleep and nutritional habits, inadequate fluid intake, medications, and stress may also increase fatigue levels.





Is there a sport that is especially safe or good for my child?

Activities that are often encouraged in our clinic include swimming, horse-back riding, yoga, Pilates, and walking. Adaptive dance and karate programs have also been found to be quite enjoyable for the child and helpful in terms of stretching and strengthening. If a child does wish to take part in activities such as long-distance running or basketball, he/she should have the appropriate running shoe or orthoses to support the feet and ankles and prevent sprains or fractures. In general, contact sports such as football are not recommended as they may increase the risk of injuries such as fractures or sprains, which may require casting and immobility.

Will schools take CMT into account when they schedule academic classes or physical education?

The answer to this question will depend on the specific needs of your child. The best way to approach these issues is to meet with the school officials and teachers and establish an IEP or Individual Educational Program. An IEP will set a plan for all the teachers involved in your child's education. An IEP is a written plan for the education of a student who has a disability or is gifted. The IEP is based on the individual student's needs and describes the special help the student will receive in school. Within Pennsylvania, you may call the Special Education ConsultLine at 1-800-879-2301. The website for the PA Department of Education is www.pde.state.pa.us/special_edu. Issues that can be addressed in an IEP include extra time for tests, use of a laptop computer, need for an aide, adaptive gym, and physical and occupational therapy. Addressing these needs before the school year starts or when entering a new school will allow the school to prepare for your child's needs. Some children may not need special adaptations to carry out activities. Others may need additional time for written work if they have hand weakness and may benefit from occupational therapy provided by the school for educational purposes.

Gym class may pose different issues. Some children have difficulty running long distances. Frequently we are asked to write a letter excusing a child from the mile run in gym class. Because we do feel exercise is important, we often ask to replace running with walking, which will allow a child to participate but not put him/her in an uncomfortable or unsafe situation. Other children may need an adaptive gym class or physical therapy instead, as they may have more difficulty taking part in activities done in a mainstream physical education program. Asking for classes to be scheduled as close to each other as possible may also help with having to walk long distances and reduce fatigue. Many schools have elevators that can be used if a child has difficulty with steps or tires during the course of the day.

AN IEP is based on the individual student's needs and describes the special help the student will receive in school.

How does CMT inheritance work?

There are many questions about the genetics of CMT which we will summarize by describing the different ways genes are passed on. There are many genes which have been found to be responsible for CMT and others still to be discovered. The following is taken from the CMTA website:

INHERITANCE PATTERNS AND CMT: Knowing which of the forms of CMT you have is important so that you can understand the implications for passing the disorder on to your children. The many different forms of CMT are inherited in different ways, so genetic counseling will vary depending on your form of CMT and its mode of inheritance. There are three distinct inheritance patterns that encompass all the many variations of CMT.

- **Autosomal dominant inheritance:** The most common forms of CMT are inherited in an autosomal dominant pattern. *Autosomal* means that the mutation occurs on a chromosome other than the X or Y chromosome. An affected person has one normal gene and one CMT gene in the relevant pair, and each child has a 50% chance of inheriting the abnormal or CMT gene and having the disorder. Boys and girls have an equal chance of inheriting the disease in this inheritance pattern. In this form, affected children can pass the CMT gene on to their children, but unaffected children do not have the abnormal gene in their DNA and cannot pass CMT to their children.

CMT is usually inherited in an autosomal dominant pattern, which means if one parent has CMT, there is a 50% chance of passing it on to each child.

- **Autosomal recessive inheritance:** The least common forms of CMT are inherited in an autosomal recessive manner. In autosomal recessive forms, both parents have to be “carriers” of the defective gene before a child can be affected. Neither parent shows signs of any symptoms of CMT, but the child who inherits autosomal recessive CMT gets a double dose of the gene, causing him/her to have the disease, usually in a more severe form. Two parents each with an autosomal recessive CMT gene have a 25% chance of passing it on to their children. It can affect both males and females. Having a recessive form of the disease may be one way to answer the question as to why a child has the disease when no one else in the family seems to have it. The parents likely do not have any symptoms and will only pass the disease on if they both carry the defective gene.
- **X-linked inheritance:** The final form of inheritance is called X-linked. In this form, the faulty CMT gene is carried on the female sex chromosome. The disease is inherited by males from their mothers, who are carriers. The mother will typically have no symptoms at all or be mildly affected. Each of her sons will have a 50% chance of having CMT, and an affected son is likely to be more severely affected than his mother. Each of the girls has a 50% chance of being a carrier. A male affected with X-linked CMT cannot pass the disorder to his sons, but all of his daughters will be carriers of the disorder. Carrier females have a 50% chance of passing the disorder to their sons. Unaffected children cannot pass the disorder onto their children because they do not carry the gene.

SPONTANEOUS MUTATION: It is also possible for CMT to develop as a result of a new or “spontaneous” mutation in the child. These cases in a family are called “de novo.” A child who has a new mutation can then pass the defective gene on to his/her children. This is another way in which a child may be affected when no one else in the family seems to have any signs of CMT.

What kind of doctor should oversee my child's health care?

Because multiple issues may arise over time, a team approach to health care is optimal. A neurologist will often be the person to first evaluate and order the diagnostic studies to make the diagnosis. A geneticist or genetic counselor may assist with questions regarding inheritance. An orthopedist will evaluate changes which may occur in the feet, hips, and spine and intervene surgically if necessary. During the teen years, when there is rapid growth, scoliosis and changes in the feet can become more prominent and need to be followed. Children and teens in our clinic are usually seen every 6 to 12 months or sooner if needed to make sure they are doing well.

What groups serve the CMT population?

Clinics in which there is a team approach with a neurologist, nurse practitioner, orthopedist, physical and occupational therapists, and orthotists often provide “one-stop shopping,” where individuals who understand CMT work together. Muscular dystrophy clinics around the country are often set up to provide such services. The Charcot-Marie-Tooth Association is an organization which can direct families to these services and answer questions for kids and their families. Camps and organizations such as the Variety Club also provide services and activities to individuals with different challenges.

How do you handle sibling rivalry between your healthy children and your child with CMT?

This too is a difficult question to answer, as sibling rivalry in and of itself is a challenge! Trying not to take sides and allowing children to work out their issues on their own may theoretically be the best way to approach the topic. Always taking sides with one child will likely not benefit either party in the long run. Trying to find common interests or activities that siblings can enjoy together may also be helpful.

What does the future hold?

Braces, wheelchairs, long-term care issues. Again, we need to take into account the individual and his/her specific needs. Some children and young adults will benefit from the use of braces or orthotics during the day to help them walk and run with less effort. Some children may only need the braces at night to stretch their heel cords. It is less likely that a child will need a wheelchair for activities of daily living. However, it is not uncommon for a child with CMT to fatigue when walking long distances. Therefore, having a wheelchair at hand may be helpful. For example, trips to the boardwalk, field trips, and Disney adventures may be times when a manual chair is helpful. Planning ahead for specific needs is important and should be addressed with your healthcare providers.

Where do we go from here?

Although CMT is not “curable” at this moment, we take the approach that it is “treatable.” That is, individuals should be cared for by providers who understand CMT and can help them address the different issues surrounding the disease. Individuals are able to lead full and productive lives. Children should be encouraged to be as active as possible and to find the activities they most enjoy. Communication with organizations such as the CMTA and Muscular Dystrophy Association (MDA) can provide valuable information about clinical trials, new medications, fundraising opportunities, and support. There is hope on the horizon as our understanding of the disease continues, and better technology is available for research and drug discovery.

THE FUTURE

for CMT patients is more promising because of the STAR initiative funded by the Charcot-Marie-Tooth Association.

To learn more about the STAR initiative, see the inside back cover of this booklet.



When We Found Out Our Daughter Had CMT

By Herb Beron, CMTA Board of Directors

In a matter-of-fact tone, the physician informed us, “The blood test came back positive—your daughter has Charcot-Marie-Tooth disorder or CMT.” These were the words told to us by our daughter’s physiatrist, a physician who specializes in physical medicine and rehabilitation.

When we first received his diagnosis, we didn’t really know what to think. What is CMT? Is it life-threatening? Can it be cured? There were so many questions. Looking back, I remember that our heads were spinning and that we didn’t know exactly what to think. On one hand we were scared and saddened to hear that our daughter Julia did indeed have a disease; maybe subconsciously we were hoping that she was just physically “slow.” On the other hand, we were indeed relieved that there was a specific concrete diagnosis for her. For us, this was of utmost importance—as we could now begin to ascend the steep learning curve about her disorder.

We voraciously gathered and read as much information about CMT as we could (both on the Internet and in the literature we received from the Charcot-Marie-Tooth Association (CMTA)). We had learned of the CMTA by simply searching the web, and they provided us with invaluable patient handbooks and vast amounts of other literature. I also consulted with an old college friend who is now a prominent orthopedist. I specifically remember him telling me, “If I had to pick one disorder for my child to have—it would be CMT.” There was something very soothing about hearing those words, which were meant to let us know that Julia could have been afflicted with a more serious neurological disease such as ALS (Lou Gehrig’s disease) or MS.

We came to learn that CMT is the most commonly inherited peripheral neuropathy, estimated to affect one in every 2,500 people, approximately 2.6 million people worldwide. CMT is a progressive disorder that causes the nerve cells leading to the body's extremities to slowly degenerate resulting in the loss of normal use of the feet/legs and hands/arms. It may also lead to a loss of sensory nerve function and deformities. CMT strikes people of all ages, genders, races, and ethnicities. The symptoms of CMT can range from relatively mild to serious, requiring a wheelchair or even a respirator. Through the CMTA, we learned of the many professionals who were leading the worldwide diagnostic and research efforts, namely Dr. Shy in Detroit and Dr. Scherer in Philadelphia. We eventually took Julia to meet Dr. Shy and his team at the CMT Clinic at Wayne State University in Detroit.

Julia had developed physically later than most children, not walking until after her second birthday. After a year of in-home physical therapy, we were strongly encouraged to take Julia to a prominent local physiatrist. We had received *so many* different opinions from our various doctors, and we really felt that we needed to put our trust in one whom we could depend on to help us “quarterback” her therapy/treatment schedule. It was this doctor who had ordered the blood work after seeing the results of her EMG. As Julia was only five at the time, it was difficult to explain the diagnosis to her, but over time she began to ask lots of questions about her disorder, and she has since been in contact with other young girls who have CMT.

My wife and I have always believed that for check-ups and garden-variety sicknesses (i.e., ear infections, colds, etc.) that our children come down with, we would use our local pediatrician. However, for more serious and more unusual issues we have always sought out the *best* specialists. The search is never an easy process. My wife and I did extensive research, spoke to countless physicians, and eventually found the specialists who had the most experience dealing with CMT, and, importantly, in whom we had the most confidence and trust. We now had to put a plan in place for Julia. Besides her semi-annual visits to her physiatrist (who was located at the same facility as her orthotist), we began a regular routine of appointments with her physical therapist, neurologist, and orthopedist—as her foot

was getting worse, and we knew that surgery was an eventuality. Julia's surgery, which comprised tendon transfer and heel cord lengthening, was performed in October, 2008. We are pleased with the results of her treatment and believe she is now well on her way to recovery. Julia now attends physical therapy 2 to 3 times per week and her progress has been outstanding. Since therapy has been a part of our family's schedule for the past several years, not a lot has changed. When she has trouble with certain tasks, Julia can get frustrated, but overall she is an incredibly well-adjusted nine-year-old with a terrific sense of who she is (and a social calendar that my wife and I have trouble keeping up with!).

One of our greatest allies during this process was and continues to be the Charcot-Marie-Tooth Association. The organization has made enormous strides in the past several years in increasing the flow of information to patients, and there is real hope that scientific breakthroughs can occur in the not-too-distant future. I would encourage all CMT patients and their families to do their best to keep up with all of this information—as the support groups, medical advisory commentary, and general news are all invaluable input.

My wife and I both firmly believe that to solve a problem and effect change, the best approach is to move to the “front lines” of the war. Last year I was elected to the CMTA Board of Directors, which has a primary goal of finding treatments and a cure for CMT, to create awareness, and to improve the quality of life for those affected by the disorder. Two years ago, we formed “TeamJulia” and joined fellow Board Member Steve O'Donnell in The Swim for the Cure, an annual swim in the Chesapeake Bay. Since then we have raised in excess of \$150,000. The other CMTA Board members and I are committed to generating the resources and funding necessary to support research to find a treatment, and ultimately a cure for CMT.

To steal a phrase from one of my daughter's favorite songs, I truly believe that “we're all in this together,” and with the work currently being done, we have the wherewithal to make a difference not just for this generation of patients, but for generations to come.



WE'RE
ALL in this
together.



How to Keep a Medical History for Your Child

Parents are an essential part of their child’s healthcare team. When medical personnel ask for vital facts, an accurate record of the child’s history of illness, medications, and surgeries can be impossible to remember, but having a written record of all those facts can literally be life-saving. For that reason, investing a little time from the beginning can make a notebook of useful medical facts a viable way to keep everyone informed about your child’s medical health. Usually, a looseleaf notebook is an easy way to start. Much like you might keep a “baby book” of important milestones and events, you simply keep a record of all your child’s pertinent medical history.

You might begin with a title page that includes the child’s name, social security number, and medical insurance information (identification number and group name, etc.). The first page would be just like the one in baby books where you list all the immunizations they have received with the dates the shots were given. If your child had any adverse reaction, list it here. The second page might list all drugs your child has been given and, again, any adverse reactions to those drugs. Also list here any allergies your child has. These could be allergies to specific drugs or seasonal allergies, or asthma. If your child has had a reaction to any medication, give the date and the nature of the reaction. Next, list all the medical problems your child is dealing with. You might begin with the diagnosis of CMT and then consider each specific problem, such as ADHD or scoliosis or any other diagnosed problem. Because you will use this notebook with nonmedical personnel as well as doctors (such as school counselors or gym teachers), you should include a brochure about CMT as well as any other simple explanations of the limitations the diagnosis places on your child’s performance. If your child takes medication on a daily basis, you should create a page

discussing the medications, the times they are taken, and the dosage strength. If your child goes to camp, is hospitalized, or just stays with friends or relatives for a weekend, this page will be invaluable. Medical treatments should be recorded next. A history of any tests that have been administered, any surgeries that have been performed, and any therapy programs your child has participated in will help medical personnel decide what to do next. With surgeries, include the date performed, the name of the procedure (tendon transfer, tendon lengthening), the name of the doctor who performed the surgery, and any other relevant information. The more information you provide, the less likelihood that tests will be duplicated for no real reason. Finally, you should have a list of all doctors who have treated your child. List their names, medical specialities, address, phone number, fax number, email address, and any other contact information. Not only is this information helpful while your child is young, but it can also follow your child all through his/her life and provide the history of care and treatment that he/she might need to recall when he/she is older and beginning his/her own family.

A medical history requires some work to assemble, but it is easy to maintain and, in an emergency, it is crucial to have accurate information readily available.

MEDICAL RECORD CHECKLIST

- Child's name, social security number, medical insurance
- Immunizations, dates
- Medications
- Allergies
- Medical diagnoses (CMT, etc.)
- History of medical procedures
- List of all doctors with contact information

Medical Terminology

Common in the Diagnosis and Treatment of CMT

Atrophy: Shrinkage or wasting away of a tissue or organ due to a reduction in its size. Atrophy is commonly caused by disease or immobilization. Nerve damage can cause muscle atrophy.

Balance: The ability to remain upright and move without falling over. Keeping one's balance is a complex process that relies on a constant flow of information about body position to the brain. Information about body position comes from three sources: the eyes; sensory nerves in the skin, muscles, and joints (called proprioceptors, which provide information about the position and movement of the different parts of the body); and the canals of the inner ear. (*Editor's Note: A person with CMT lacks proprioception [defined on page 25] because of sensory loss.*)

Bilateral: Affecting both sides of the body.

Brace, orthopaedic: An appliance worn to support part of the body or hold it in a fixed position. A brace may also help the movement of a limb when movement would otherwise be impossible. A person who has lost the ability to flex the foot upward and drags the toes on the ground with each step can be fitted with a device called a footdrop splint that keeps the foot permanently at right angles to the leg and thus allows walking.

KNOWING

the medical “language”
makes doctors’ visits
less intimidating.

Brachial plexus: A collection of nerves that pass from the lower part of the spine and the upper part of the thoracic spine down the arm. This collection of nerves controls the muscles in, and receives stimulation from, the arm and hand.

Brachialgia: Pain or stiffness in the arm. It is often accompanied by pain, tingling, or numbness of the hands or fingers and weakness of hand grip.

Calcaneus: The heel bone. The Achilles tendon is fixed to the back of the heel bone and controls the up and down movement of the foot.

Central nervous system: The anatomical term for the brain and spinal cord, often abbreviated CNS. The central nervous system works in tandem with the peripheral nervous system (PNS), which consists of all the nerves that carry signals between the CNS and the rest of the body.

Chromosomes: Threadlike structures present within the nuclei of cells. Chromosomes carry the inherited genetic information that directs the activities of cells and, thus, the growth and functioning of the entire body.

Clawfoot: A deformity of the foot that includes an exaggerated arch and turning under of the tips of the toes.

Clawhand: A deformity of the hand in which the fingers are permanently curled.

Contracture: A deformity caused by shrinkage of scar tissue in the skin or connective tissue, or by irreversible shortening of muscles and tendons.

Demyelination: Breakdown of the fatty shields that surround and electrically insulate nerve fibers. The sheaths provide nutrients to the nerve fibers and are vital to the passage of electrical impulses along them. Demyelination “short-circuits” the functioning of the nerve, causing loss of sensation, coordination, and power in specific areas of the body.

Distal: Describing part of the body that is farther away from a central point of reference, such as the trunk of the body. The fingers are distal to the arm. The opposite of distal is proximal.

DNA: The commonly used abbreviation for deoxyribonucleic acid, the principle carrier of genetic information in almost all organisms. DNA is found in the chromosomes of cells.

Dorsum: The top aspect of the foot. The opposite of dorsal is ventral or anterior.

EMG: The abbreviation for electromyogram, a test in which the electrical activity in the muscle is analyzed after being amplified, displayed, and recorded. An EMG is done to reveal the presence of muscle disorders or disorders in which the nerve supply to the muscle is impaired.

Exacerbation: A worsening. (*Editor's Note: Neurotoxic drugs can cause an exacerbation of CMT.*)

Footdrop: A condition in which the foot cannot be raised properly. It hangs limp from the ankle joint, causing it to catch on the ground when walking.

Gait: The style or manner of walking. Some neuromuscular disorders are evaluated on the basis of altered gait (i.e., CMT).

Gene: A unit of material of heredity. A gene consists of a short section of DNA contained within the nucleus of a cell. It influences the specific workings of a cell; the activity of the same gene in many different cells specifies a particular physical or biochemical feature of the whole body.

Genetics: The study of inheritance, i.e., how the characteristics of living organisms are passed from one generation to another.

Hallux: The medical term for the big toe.

Hallux rigidus: Loss of movement in the large joint at the base of the big toe.

Hallux valgus: A deformity of the big toe in which the joint at the base projects outward and the top of the toe turns inward. A hallux valgus often results in a bunion (a firm, fluid-filled swelling over the joint).

Hammer toe: A deformity of the toe in which the main toe joint is bent upward like a claw. The deformity is caused by an abnormality of the tendons in the toe.

Idiopathic: Of unknown cause.

Kyphosis: The term for excessive backward curvature of the spine. Kyphosis usually affects the spine at the top of the back, resulting in either a hump or a more gradually rounded back.

Median nerve: A branch of the brachial plexus; one of the main nerves that runs down the arm's full length into the hand. The median nerve controls the muscles of the forearm and hand. This nerve also conveys sensations from the thumb, index finger, middle finger, part of the ring finger, and the palm at the base of those fingers.

Myelin: The fatty material composed of lipids and protein that forms a protective sheath around some types of nerve fiber. Myelin acts as an electrical insulator, increasing the efficiency of nerve impulse conduction. Abnormal breakdown of myelin is called demyelination.

Neuropathy: Disease, inflammation, or damage to the peripheral nerves which connect the central nervous system to the sense organs, muscles, glands, and internal organs. Symptoms include numbness, tingling, pain, or muscle weakness.

Neurotoxin: A chemical which damages nervous tissue. The principal effects of neurotoxic nerve damage are numbness, weakness, or paralysis of the part of the body supplied by the affected nerve.

Occupational therapy: Treatment aimed at enabling people disabled by physical illness to relearn muscular control and coordination, to cope with everyday tasks and, if possible, to resume employment.

Osteotomy: An operation in which a bone is cut to change its alignment or to shorten or lengthen it.

Paraparesis: Partial paralysis or weakness of both legs and sometimes, part of the trunk.

Paresis: Partial paralysis or weakness of one or several muscles.

Paresthesia: Altered sensation in the skin that causes numbness or tingling. (Pins and needles syndrome.)

Patella: The medical name for the kneecap. The patella is held in place by the quadriceps muscle at the front of the thigh.

Peripheral nervous system: All the nerves that fan out from the central nervous system (brain and spinal cord) to the muscles, skin, internal organs, and glands. Diseases and disorders affecting the peripheral nerves are grouped under the term *neuropathy*.

Peroneal muscular atrophy: An inherited disorder characterized by wasting of the muscles, first in the feet and legs and then in the hands and forearms. The condition, also known as Charcot-Marie-Tooth disorder, is a result of degeneration of some of the peripheral nerves or of the myelin sheath which surrounds them.

Pes cavus: The medical term for clawfoot.

Phrenic nerve: The principal nerve supplying the diaphragm. It carries all of the motor impulses to, and some of the sensory impulses from, the diaphragm, and plays an important part in controlling breathing.

Proprioception: The body's internal system for collecting information about its position relative to the outside world and the state of contraction of its muscles (i.e., maintaining balance). This is achieved by means of sensory nerve endings within the muscles, tendons, joints, and cells in the balance organ of the inner ear. Information from these sensory nerve endings helps the muscles contract so that balance and posture are maintained.

Proximal: Describing a part of the body that is nearer to a central point of reference, such as the trunk of the body. The opposite of proximal is distal.

Scoliosis: A deformity in which the spine is bent to one side. The chest and the lower back are most commonly affected. Scoliosis usually starts in childhood or adolescence and progressively becomes more marked until the age at which growth stops.

Supination: The act of turning the body to a supine (lying on the back with the face upward) position or the hand to a palm forward position. Movements in the opposite direction are called pronation.

Tendon: A fibrous cord that joins muscle to bone or muscle to other muscle.

Tendon transfer: An operation to reposition a tendon so that it causes a muscle to perform a different function. Tendon transfer may be used to restore function impaired by a deformity such as clubfoot, paralysis, or thumb movement.

Tremor: An involuntary, rhythmic, oscillating movement of the muscles of part of the body, most often the hands, feet, jaw, tongue, or head. Tremor is caused by rapidly alternating contraction and relaxation of the muscles.

Varus: The medical term for an inward deformity of a part of the body. For example, in genu varum (bowleg), the lower leg is displaced inward.

Wristdrop: Inability to straighten the wrist so that the back of the hand cannot be brought horizontal with the back of the forearm. This causes weakness of grip because the hand muscles can function efficiently only when the wrist is held straight.

X-linked disorders: Sex-linked genetic disorders in which the abnormal gene or genes are located on the X chromosome, and in which almost all those affected are male.



Resources for CMT Patients and Family Members

Many people diagnosed with CMT feel as though they are all alone in dealing with their disorder. That should not be the case. There are several direct service providers who can help in providing proper medical care. There is a diagnostic DNA testing service available for CMT. There is information provided by non-profit organizations, such as the CMTA, and there are forums and chat rooms where even the most isolated person can connect with someone else who has CMT.

Here is the information necessary to connect with the people and organizations who can help.

DIRECT PATIENT CARE

In the direct patient care category, there are two well-known charitable organizations.

The Muscular Dystrophy Association

The Muscular Dystrophy Association (MDA) operates approximately 200 hospital-affiliated clinics across the country and serves CMT patients of all ages. In MDA clinics, a person receives diagnostic and follow-up care from specialists in neuromuscular diseases and can receive assistance with the purchase and repair of wheelchairs and leg braces.

The only requirement for receiving an evaluation at an MDA clinic is the written recommendation of a physician who has determined the person may have one of the neuromuscular diseases covered by the MDA's medical services program. (CMT is one such disease.)

MDA pays for only those services authorized in its program that are not covered by private or public insurance plans. Their payments are made directly to the institution in which the MDA clinic is located or to authorized vendors. No services or durable medical equipment may be ordered directly by the patient or their family if they wish the MDA to pay for the item or service.

In addition to the preliminary diagnostic process, the MDA clinics provide invaluable management care. They schedule annual follow-up visits and may prescribe physical therapy, occupational therapy or respiratory therapy (one consultation annually to evaluate the need and to instruct the patient or family in continuing therapy), social services to help patients find additional resources for payment of medical services, cooperation with the person's personal physician in the form of summary reports, genetic counseling, support groups and flu inoculations. The MDA also sponsors summer camps for youngsters.

In those cases where family or community resources are not available, the MDA assists in arranging transportation to appointments at the nearest MDA clinic. For complete information about the services of the MDA, go to their home page at www.mda.org or call 1-800-344-4863.

Shriners Hospitals for Children

Any child may be eligible for care at Shriners Hospitals if the child is under the age of 18 and there is a reasonable possibility the child's condition can be helped. There is never a charge to the patient or the parents for any medical care or services. For more information about Shriners Hospitals, visit www.shrinershq.org/Hospitals/_Hospitals_for_Children/. Application forms for admission to the orthopedic hospitals can be downloaded in Adobe format or obtained by calling the toll-free referral line at 1-800-237-5055. (In Canada, call 1-800-361-7256.) The hospitals offer a full spectrum of orthopedic care, including bracing, therapy, and corrective surgery.

SHRINERS HOSPITALS

Orthopedic Locations:

Boston, MA	Los Angeles, CA	St. Louis, MO	<i>Shriners International Headquarters is in Tampa, Florida, and can be reached by phoning 1-813-281-0300.</i>
Chicago, IL	Mexico City, Mexico	Salt Lake City, UT	
Erie, PA	Minneapolis, MN	Shreveport, LA	
Greenville, SC	Montreal, QC, Canada	Spokane, WA	
Honolulu, HI	Philadelphia, PA	Springfield, MA	
Houston, TX	Portland, OR	Tampa, FL	
Lexington, KY	Sacramento, CA		

CMT CLINICS

Wayne State Clinic

The clinic at Wayne State was founded in 1977 and has seen patients from the United States and 12 other countries since its inception. In addition to providing clinical care, they also use the information gleaned from patient evaluations to add to their clinical research studies on the natural history and progression of CMT and related conditions.

Initially a patient meets with a genetic counselor to talk about the study and the specific testing that will be done. Genetic issues related to CMT and options for genetic testing are discussed. Tests that are done include sensory testing, strength testing, nerve conduction velocity testing, and hand function testing. A neurologist will evaluate each patient by reviewing his/her medical history and doing a neurological exam. A physiatrist will address possible rehabilitative needs such as bracing or physical and occupational therapy.

To make an appointment or to ask specific questions about the clinic, call 1-313-577-1689.

The John P. Murtha Neuroscience and Pain Institute

The Charcot-Marie-Tooth program at the John P. Murtha Neuroscience and Pain Institute was begun in August of 2003. The CMT specialists there take a team approach to CMT diagnosis. At the first visit, they take a medical history and a family history. Diagnostic tests such as neurological and physical exams, nerve conduction studies, electromyography, and genetic testing may be done. One of the offerings of the clinic is a six-week program called "Healthy Living with a Chronic Condition."

A patient can refer him/herself or be referred by a doctor. For more information, call 1-877-576-5700.

Hospital of the University of Pennsylvania (HUP)

The Children's Hospital of Philadelphia (CHOP)

The Hospital of the University of Pennsylvania and the Children's Hospital of Philadelphia are CMTA Centers of Excellence. There are six such centers throughout the US which will serve as repositories of patient information and will use the standardized CMT Neuropathy Score to evaluate the level of disability a patient is experiencing. Once medications for CMT are available, these centers will serve as clinical trial headquarters.

For more information about HUP's CMTA program, contact Steven S. Scherer, MD, PhD at 215-349-5313. For more information about CHOP's CMTA program, contact Richard S. Finkel, MD at 215-590-1719.

GENETIC TESTING AND COUNSELING

Genetic diagnosis of CMT is important when families are considering inheritance probabilities for future children and when the more traditional measurements, such as nerve conduction velocities and electromyograms are inconclusive. Comprehensive genetic diagnostic testing is available from one commercial company: Athena Diagnostics, in Worcester, MA. Information about the testing procedure and the possible costs can be obtained by calling customer service at 1-800-393-4493, or by visiting www.athenadiagnostics.com. The tests must be ordered by a physician, but information can be gathered by patients before making any decisions regarding the tests. Currently, Athena offers tests for 1A, 1B, 1C, 1D (ERG 2), 1E, 1F, 1X, 2A, 2E, 2I, 2J, 2K, 4A, 4E, 4F, HNPP, CHN, and DSN; however, the list is always growing.

In addition to the genetic testing, Athena also offers genetic counseling and test interpretation to physicians so that they can better serve their patients once the test information has been received.

Independent genetic counselors may also be located through the National Society of Genetic Counselors. Visit www.nsgc.org or call 1-312-321-6834.

THE CMTA

can provide a wealth
of information for the
newly diagnosed.

ORGANIZATIONS, DISCUSSION FORUMS, AND OTHER RESOURCES

The final source of help and information for patients and families dealing with CMT (and some would say the most important) are the organizations and groups that provide support and information on living with the disorder.

The Charcot-Marie-Tooth Association

2700 Chestnut Street
Chester, PA 19013
1-800-606-2682
www.charcot-marie-tooth.org or www.cmtinfo.org.

Founded in 1983, this organization publishes a newsletter six times a year, hosts patient/family conferences, funds numerous CMT research grants, and has published books and pamphlets about the disorder and how to deal with it.

ORGANIZATIONS, DISCUSSION FORUMS, AND OTHER RESOURCES (continued)

Charcot-Marie-Tooth Association Australia, Inc.

www.e-bility.com/cmtaa

CMT Family Support

http://health.groups.yahoo.com/group/CMT_Family_Support

Info, articles, and a discussion forum.

CMT United Kingdom

www.cmt.org.uk

A new and useful information source from England.

CMTUS

<http://health.groups.yahoo.com/group/CMTUS>

Information, research articles, and a discussion forum.

Hereditary Neuropathy with Liability to Pressure Palsies (HNPP)

www.hnpp.org

Information and resources related to HNPP.

Muscular Dystrophy Association

1-800-344-4863

www.mda.org

CMT is one of the MDA's 40 diseases. For more information about the MDA, see page 28.

Additional resources may be found on the Resources page of the CMTA website at <http://www.charcot-marie-tooth.org/resources.php>. These include state-by-state lists of physicians familiar with CMT, support groups, and other agencies and organizations that provide information and assistance.

STAR... the promise of a future without CMT

In 2008, the CMTA launched the Strategy to Accelerate Research (STAR) as a three-step strategic research program to maximize breakthroughs in genetics and dramatically speed up the pace of CMT research.

The goals of STAR were ambitious, but doable:

- 1) within 5 years, introduce effective therapies for the three most common types of CMT
- 2) within 10 years, reverse the symptoms of the disorder

Gifts directed to STAR will be used exclusively to fund this ambitious research program. Send donations to CMTA STAR, 2700 Chestnut St., Chester, PA, 19013. Call 1-800-606-2682 for credit card gifts or go online to www.cmtausa.org/STAR. Deductions are 100% tax-deductible.



CMT is the most common
inherited peripheral neuropathy.
Worldwide, more than
2.6 million people have CMT.



*Published by the Charcot-Marie-Tooth Association
with a grant from the PA Department of Health.*

Edited by Pat Dreibelbis, Director of Program Services, CMTA.