Dear Friends:

I’ve lived with Charcot-Marie-Tooth disease (CMT) since my early 20s — more than half my life. The disease has progressed slowly over the years, mostly affecting my lower legs and hands, so that now I use a manual wheelchair part time. In those years, I’ve continued a career in computer technology, started a small business, pursued my interests in art and photography, married, and contributed my knowledge and experience to others with disabilities.

This booklet has been prepared to give you the basic knowledge about CMT and Dejerine-Sottas disease (DS) that you’ll need in order to help you prepare for changes that may occur in your future. You’ll learn that CMT is usually quite slow in progression and that, while it presents challenges in daily life, there are many techniques and devices to help you adapt to those challenges.

You’ll read that many different genetic causes of CMT have been found, and cases vary greatly. But CMT is almost never life-threatening, and it seldom affects the heart and breathing functions.

And it doesn’t affect intelligence or the spirit. I know of many productive, successful people with CMT — doctors and scientists, artists and singers, athletes and teachers, active teens and students. I know children with CMT who have bright futures. We’ve all learned to strike a balance between adapting to our limitations and surroundings, and living a fulfilling life despite them.

I have the wonderful support of my family and great friends. I’m involved in volunteer projects that help young people with disabilities, advising them on independence and entrepreneurship, and raising public awareness about disability wherever I go. What I’ve learned — and what I try to teach — is that people with disabilities are — like everyone else — full of possibilities and gifts. These, not our limitations, are what matter.

Another important extended family in my life is the Muscular Dystrophy Association, which offers a great program of services, leads the world in CMT research and keeps us well informed about the disease. See “MDA Is Here to Help You,” on page 12, for details of the Association’s program.

While MDA’s research program continues making strides toward better treatments and a cure, it’s good to know that people with disabilities have more opportunities than ever before to develop and use their abilities, and that the laws entitle us to equal employment opportunities and access to public places.

As you face the challenges ahead, remember, MDA and all its resources are there to help you and your family. You’re not alone.

George J. Donahue
Watertown, Massachusetts
Charcot-Marie-Tooth disease (CMT) is a neurological disorder, named after the three physicians who first described it in 1886 — Jean-Martin Charcot and Pierre Marie of France, and Howard Henry Tooth of the United Kingdom. Although most people have never heard of CMT, it affects some 115,000 Americans.

Unlike other neurological disorders, CMT usually isn’t life-threatening, and it almost never affects the brain. It causes damage to the peripheral nerves — tracts of nerve cell fibers that connect the brain and spinal cord to muscles and sensory organs.

Peripheral nerves control movement by relaying impulses from the spinal cord to muscles. They convey sensation by carrying feelings like pain and temperature from the hands and feet to the spinal cord. They also help control balance, by carrying information about the position of the body in space. They transmit information about the feet and hands to the spinal cord and then the brain, so that the brain knows where to place the feet when walking and where the hands should be placed to reach for something.

Nerve damage, or neuropathy, causes muscle weakness and wasting, and some loss of sensation, mostly in the extremities of the body: the feet, the lower legs, the hands and the forearms.

Although CMT can look very similar to an acquired neuropathy — a type of nerve damage caused by diabetes, immunological abnormalities or exposure to certain chemicals or drugs — it isn’t caused by anything a person does, and it isn’t contagious. It’s hereditary, meaning that it can be passed down through a family from one generation to the next. (See “Does It Run in the Family?” page 10.)

Because of these features, CMT is sometimes called hereditary motor and sensory neuropathy (HMSN). Some doctors also use the old-fashioned name peroneal muscular atrophy, which refers to wasting of the peroneal muscle in the lower leg.

There are even more names for CMT because the disease exists in many different forms, each unique in its severity, age of onset, progression and exact symptoms. For example, Dejerine-Sottas disease (DS) is a severe form of CMT that manifests during infancy or early childhood.

Although there’s no cure for CMT, there are treatments that can be used to effectively manage its symptoms. Those treatments, described here along with a general overview of CMT, have allowed many people with the disease to lead active, productive lives.

What causes CMT?

CMT is caused by defects in genes, which are segments of DNA contained in the chromosomes of the body’s cells. Genes are recipes for making the proteins that serve essential functions in our bodies. Each form of CMT is linked to a specific gene, and all of those genes make proteins found within the peripheral nerves.

Peripheral nerves provide an essential relay between your brain and the rest of your body. When you decide to move your leg, your brain sends an electrical signal to muscle-controlling nerve cells in your spinal cord, which then use the peripheral nerves to pass the signal on to your leg muscles.

And if you hurt your leg, you feel it because pain-sensitive nerve cells there

What Is Charcot-Marie-Tooth Disease?

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And if you hurt your leg, you feel it because pain-sensitive nerve cells there
have sent a signal through your peripheral nerves to your brain.

The peripheral nerves are made up of fibers, or axons, that extend from sensory nerve cells and muscle-controlling nerve cells, and carry electrical signals to and from the spinal cord.

In order for you to move and react with precision and speed, axons have to transmit their signals within a fraction of a second. This is a real challenge for axons that have to stretch over long distances, like the ones connected to muscles in your fingers and toes.

To give axons a performance boost, each one is surrounded by a coating called myelin. Similar to the way plastic coating is used to insulate electrical wiring, myelin insulates the electrical signals in axons. It also provides essential nourishment to the axons.

Some 20 genes have been implicated in CMT, each one linked to a specific type (and in many cases, more than one type) of the disease. (See “What are the different types of CMT?” page 7.) Some of those genes make proteins needed in axons, and others make proteins needed in myelin.

Defective myelin genes can cause a breakdown of myelin (called demyelination) while defective axon genes can cause an impairment of axon function (axonopathy).

In either case, the end result is the same: Defects in the axon or the myelin cause progressive damage to the axons.

The longest axons in the body are especially sensitive to damage, which explains why CMT mostly causes motor and sensory problems in the body’s extremities.

Nerves other than those that go to and from the extremities can be affected at the severe end of the CMT spectrum. If the nerves that go to and from the diaphragm or intercostal (between the ribs) muscles are affected, respiratory impairment can result.

What happens to someone with CMT, and how is it treated?

Partly because there are different types of CMT, the exact symptoms vary greatly from person to person. This section provides a general picture of CMT, and the next section describes different types of the disease.

Muscle weakness

In general, people with CMT experience slowly progressive weakness and wasting in the distal muscles, which control the extremities. These muscles control foot and hand movements. More proximal muscles, those closer to the trunk, such as the leg and arm muscles, are rarely affected.

Usually, weakness begins in the feet and ankles, and manifests itself as foot drop — difficulty lifting the foot at the ankle, so that the toes point downward during walking. Foot drop causes frequent tripping, and with increasing weakness and attempts at compensation, the affected person develops an abnormal gait.

Many people with CMT make their first visits to a neurologist after they notice frequent trips and falls, ankle sprains, or ankle fractures, caused by foot drop.

When these problems occur, some people find they can overcome them just by wearing boots or high-top shoes to support the ankles.

Others might require leg braces, such as an ankle-foot orthosis (AFO), a
you should have your breathing checked by a specialist, who might recommend occasional or nighttime use of a device that delivers air under pressure into the lungs.

Although it’s usually too slight to cause disability or discomfort, some people with CMT experience tremor (involuntary shaking). CMT with obvious tremor is sometimes called Roussy-Levy syndrome.

**Contractures and bone deformities**

Many people with CMT eventually develop contractures (stiffened joints) that result in deformities of the feet and hands.

The contractures occur because as some muscles around a joint weaken, others remain strong, contracting and pulling on the joint. Over time, the bones around the joint shift into abnormal positions.

For example, as muscles that lift the foot at the ankle become weak, muscles that lower and curl the foot downward contract and tighten, causing the most common type of foot deformity — a shortened foot with a high arch (pes cavus). As the contracture gets worse, the toes can become locked in a flexed position.

A small fraction of people with CMT develop “flat feet” (pes planus), presumably because of a different pattern of muscle weakness.

During walking, these deformities can cause unusual friction against the toes, heel and ball of the foot, leading to painful abrasions, blisters and calluses. If left untreated, the contractures and secondary abrasions tend to worsen over time, making it increasingly difficult to walk.

As CMT progresses, contractures in the hand can lock the fingers in a flexed position, and in rare cases severe proximal weakness can lead to scoliosis (side-to-side curvature of the spine) or kyphosis (front-to-back spine curvature).
Paradoxically, some people with CMT experience more pain — a combination of painful muscle cramps and neuropathic pain. This pain isn’t caused by an external trigger, but by defective signals in sensory axons. Both types of pain usually can be alleviated with medication.

In many people with CMT, sensory loss is associated with dry skin and hair loss in the affected area.

In rare cases, sensory loss can include gradual hearing impairment and sometimes deafness. Watching out for these potential problems will enable you to seek appropriate treatment if necessary.

**Drug warning**

The use of certain prescription drugs or excess alcohol can lead to acquired neuropathy, and thus might exacerbate CMT. Case studies have shown that the chemotherapy drug vincristine can cause rapid deterioration in people with CMT.

When taking a prescription drug for the first time, it’s a good idea to consult your doctor about its possible effects on CMT. Or, enter the specific name of the drug into an Internet search engine, along with the words “prescribing information,” to receive a full explanation of what the drug does and what its side effects may be.

You’re unlikely to see anything specific about CMT. However, if the medication’s side effect description mentions words like neuropathy, paresthesias, neuropathic pain or peripheral nerve damage, you may want to consult your physician about its use in CMT and possible alternatives.

Lists of contraindicated (forbidden) drugs for people with CMT are often composed mostly of medications used to treat serious conditions, such as cancer. In these cases, there may be no alternative to taking the drug, with the awareness that CMT symptoms may worsen.

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A small fraction of people with severe CMT also experience hip displacement at an early age.

One of the most effective ways to keep muscles from tightening up and forming contractures is to begin a regular program of physical therapy, which usually consists of low-impact exercises and stretching.

Your MDA clinic can help get you started on an individualized physical therapy program.

Foot contractures also can be delayed by using AFOs, which force the feet into a normal position and decrease stress on the ankles. Similarly, splints can be used to prevent unintended flexing of the toes and fingers.

If these methods fail and severe contractures occur, surgery can be used to loosen up tight muscles and tendons, or to correct bone deformities. Surgery is often necessary for advanced scoliosis.

**Sensory loss and associated symptoms**

Because CMT causes damage to sensory axons, most people with CMT have a decreased sensitivity to heat, touch and pain in the feet and lower legs.

Although people with CMT often complain that their feet get cold (caused as much by a loss of insulating muscle as by damage to sensory axons), most of these sensory losses are undetectable except by a neurological exam — but it’s important to recognize that they occur.

Combined with the regular abrasions caused by foot deformities (see page 5), the lack of pain sensitivity makes people with CMT at risk for developing ulcerations — wounds that have gone unnoticed and become severely infected. If you have CMT, and especially if you have any foot deformities, you should check your feet regularly for injuries.
What are the different types of CMT?

The many different types of CMT are distinguished by age of onset, inheritance pattern, severity, and whether they’re linked to defects in axon or myelin.

While those distinctions are useful, it’s important to realize that, because of the vast number of genetic defects that can lead to CMT, some people fall on the borders between different types and many have specific “subtypes” not detailed here.

(For more information about the genetics and inheritance of CMT, see “Does it Run in the Family?” page 10.)

CMT1 and CMT2

Onset:
usually childhood or adolescence

Inheritance:
type 1, autosomal dominant; type 2, autosomal dominant or recessive

Features:
These are the two most common forms of CMT. (In fact, a subtype called CMT1A, caused by a defect in the PMP22 gene on chromosome 17, accounts for around 60 percent of all CMT cases.)

CMT1 is caused by demyelination, and CMT2 is caused by axonopathy, but both produce the classic symptoms described above.

CMT2 is sometimes associated with a treatable condition called restless legs syndrome, an irresistible urge to move the legs while sitting or lying down.

CMTX

Onset:
childhood or adolescence

Inheritance:
X-linked

Features:
CMTX has symptoms similar to those of CMT1 and CMT2. Because of its linkage to the X chromosome, it often affects males more severely than females.

CMT4

Onset:
infancy, childhood or adolescence

Inheritance:
autosomal recessive

Features:
CMT4, a demyelinating form of CMT, causes weakness, usually mostly distal, but sometimes involving proximal muscles. Sensory dysfunction can also occur. When CMT4 begins in infancy, it’s characterized by low muscle tone. Young children with CMT4 generally have delayed motor (movement-related) development.

Dejerine-Sottas disease

Onset:
ear early childhood (generally before 3 years)

Inheritance:
autosomal dominant or recessive

Features:
DS is sometimes classified as a subgroup of CMT4 and is also sometimes called HMSN3. It’s a severe neuropathy, with generalized weakness sometimes progressing to severe disability, loss of sensation, curvature of the spine and sometimes mild hearing loss.

Several of the genes that, when flawed, cause Dejerine-Sottas disease, are the same genes that, when flawed in a different way, lead to various forms of CMT.

For axons and Schwann cells, communication is the key to a healthy relationship. Axons send chemical messages that attract Schwann cells and encourage myelin formation, and Schwann cells appear to send messages that nourish and protect axons. The various genetic defects that cause CMT often disrupt these interactions.
Congenital hypomyelinating neuropathy (CHN)

Onset:
congenital (at or near birth)

Inheritance:
autosomal recessive, spontaneous

Features:
Unlike other types of CMT, CHN is associated with reduced myelin formation (hypomyelination) from birth rather than a breakdown of existing myelin. Both genetically and clinically, it’s similar to DS, but usually has an earlier onset and a nonprogressive or slowly progressive course.

Many children with CHN grow up and experience gradual improvements in strength.

How is CMT diagnosed?

A combination of lower leg weakness and foot deformities is a red flag for CMT, but isn’t sufficient for diagnosis. When a patient has those symptoms, a well-trained neurologist will usually start with a physical exam to look for further signs of distal weakness and sensory loss.

As a test for leg weakness, the neurologist might ask the patient to walk on his heels, or to move part of his leg against an opposing force.

To look for sensory loss, the neurologist will usually test the patient’s deep tendon reflexes (like the knee-jerk reflex), which are reduced or absent in most people with CMT.

During this initial evaluation, the neurologist also will ask about the patient’s family history. A family history of CMT-like symptoms, combined with signs of nerve damage from the individual’s physical exam, strongly point to CMT or another hereditary neuropathy.

Lack of a family history doesn’t rule out CMT, but might prompt the neurologist to ask about diabetes, overexposure to certain drugs and other potential causes of neuropathy.

Axons, Myelin and CMT

There are more than 30 genes that, when flawed, can cause CMT or Dejerine-Sottas disease. Many carry instructions for structural components of the axon (nerve fiber) or of the myelin sheath that surrounds it.

Myelin structural components

Peripheral myelin protein 22 (PMP22) — CMT1(A), DS*, CMT4
Controls Schwann cell division.

Myelin protein zero (MPZ, or P0) — CMT1(B), CMT2, DS*, CMT4
Holds layers of myelin together.

Connexin 32 (Cx32, a.k.a. GJB1) — CMTX
Forms pores between layers of myelin.

Axon structural components

Neurofilament-light (NF-L) — CMT2 (single large Russian family)
Acts as backbone and conveyor belt within axon.

*DS = Dejerine-Sottas
Next, if the diagnosis is still consistent with CMT, the neurologist may arrange for genetic testing. These tests, done by drawing a blood sample, are designed to detect the most common genetic defects known to cause CMT. Many, but certainly not all, of the genetic mutations underlying CMT can be detected with a DNA blood test.

A positive genetic test result can provide a definite diagnosis and useful information for family planning. But once again, a negative result doesn’t rule out CMT.

The neurologist also may perform a nerve conduction velocity (NCV) test, which measures the strength and speed of electrical signals transmitted through nerves.

It’s done by placing surface electrodes, similar to those used for electrocardiograms, on the skin at various points over a nerve. One electrode delivers a mild shock that stimulates an electrical response in the nerve, and the others record this response as it travels through the nerve.

Delayed responses are a sign of demyelination and small responses are a sign of axonopathy. Thus, NCV is often used to distinguish between CMT1 and CMT2.

Other procedures sometimes used to diagnose CMT include electromyography (EMG), which measures the electrical signals in muscles, and less commonly, nerve biopsy, which involves the removal and examination of a small piece of nerve.
CMT can run in a family, even when there’s no obvious family history of it. In part, this is because CMT can be inherited in three different ways that aren’t always easy to trace through a family tree: *X-linked, autosomal dominant and autosomal recessive.*

*X-linked* means that the genetic defect (or *mutation*) is located on the X chromosome. In females, who have two X chromosomes, a normal copy of the gene on one chromosome can often compensate (at least partially) for the defective copy. Therefore, X-linked diseases usually affect males more severely than females, because males only have one X chromosome. X-linked diseases (like CMTX) can’t be passed from father to son.

*Autosomal* means the mutation occurs on a chromosome other than the X or Y. Therefore, *autosomal diseases* affect males and females equally. *Autosomal recessive* means that two copies of a defective gene are required for the full-blown disease. One copy is inherited from each parent, neither of whom would normally have the disease. *Autosomal dominant* means one copy of a defective gene is enough to cause disease. A person who inherits the defective gene from a parent will have the disease, as will the parent.

When CMT is passed on in an autosomal dominant pattern, it can be easy to recognize in the family tree. In contrast, X-linked or autosomal recessive types of CMT might seem to occur “out of the blue.” But in reality, the mother or both parents might be carriers who silently harbor a genetic mutation. Many parents have no idea they’re carriers of a disease until they have a child with the disease.

CMT also can occur when a new mutation occurs during the child’s conception. These are called *spontaneous mutations,* and after they occur, they can be passed on to the next generation.

Your risk of inheriting or passing on CMT depends largely on what type of CMT you have (see “What are the different types of CMT?” page 7.) A good way to find out more about this risk is to talk to your MDA clinic physician or a genetic counselor at the MDA clinic. Also, see MDA’s booklet “Facts About Genetics and Neuromuscular Diseases.”
In 1991, the genetic causes of CMT were completely unknown. But just 10 years later, MDA-funded scientists had helped to identify 10 CMT-linked genes and found evidence for several others. (There are now thought to be some 30 genes in which flaws can cause CMT.) This accomplishment has led to genetic testing for many types of CMT, which has greatly improved diagnosis.

Of equal importance, the ongoing hunt for CMT genes has given insights into treatments that might be used to stop or reverse the disorder. As the CMT gene hunt continues, MDA-funded scientists are investigating how and why specific genetic mutations lead to different types of CMT. These insights are expected to lead to improved ability to predict the course of CMT in specific individuals and ultimately to lead to treatments.

In addition, MDA-funded scientists have made significant progress in understanding the biology of axons and Schwann cells — the cells that make myelin in the peripheral nerves. The goal is to restore normal myelin maintenance.

One scientist is conducting laboratory experiments to see whether a compound called heat shock protein 90 might be therapeutic in the type 1A form of CMT, while others are conducting an MDA-funded clinical trial to test the effects of high-dose ascorbic acid on CMT1A.

In addition to these specific projects, MDA is supporting the CMT North American Database, a secure repository of information about CMT provided by patients and families. (See http://neurology.med.wayne.edu/neurogenetics/na_database.php.) MDA also is supporting the North American CMT Network, an extension to the Database designed to provide an infrastructure for CMT research.
MDA Is Here to Help You

The Muscular Dystrophy Association offers a vast array of services to help you and your family deal with CMT. The staff at your local MDA office is there to assist you in many ways. The Association’s services include:

- nationwide network of clinics staffed by top neuromuscular disease specialists
- MDA summer camps for kids with neuromuscular diseases
- help with locating durable medical equipment through its national equipment program
- financial assistance with repairs or modifications to all types of durable medical equipment
- annual occupational, physical, respiratory or speech therapy consultations
- annual flu shots
- support groups for those affected, spouses, parents or other caregivers
- online support services through the e-community myMDA and through myMuscleTeam, a program that helps recruit and coordinate in-home help

MDA’s public health education program helps you stay abreast of research news, medical findings and disability information through magazines, publications, educational speakers, seminars, videos and newsletters.

MDA’s website at www.mda.org contains thousands of pages of valuable information, including disease specifics, research findings, clinical trials and past magazine articles.

Everyone registered with MDA automatically receives Quest, MDA’s award-winning quarterly magazine. Quest publishes detailed articles about research findings, medical and day-to-day care, helpful products and devices, social and family issues, and much more. Other MDA publications can be found at www.mda.org/publications; many booklets are available in Spanish. Ask your local office for “MDA Services for the Individual, Family and Community” and for help with obtaining copies of other publications.

If you have any questions about CMT, someone at MDA will help you find the answer. To reach your local MDA office, call (800) 572-1717.

On the cover:
TyKiah has been affected by CMT since childhood. She earned bachelor's and M.B.A. degrees from Wright State University in Dayton. In 2001, she established the WrightChoice Intern Program, which helps minority students and students with disabilities find internships and jobs.
**MDA’s Purpose and Programs**

The Muscular Dystrophy Association fights neuromuscular diseases through an unparalleled worldwide research effort. The following diseases are included in MDA’s program:

**Muscular Dystrophies**
- Myotonic dystrophy (*Steinert disease*)
- Duchenne muscular dystrophy
- Becker muscular dystrophy
- Limb-girdle muscular dystrophy
- Facioscapulohumeral muscular dystrophy
- Congenital muscular dystrophy
- Ocularpharyngeal muscular dystrophy
- Distal muscular dystrophy
- Emery-Dreifuss muscular dystrophy

**Motor Neuron Diseases**
- Amyotrophic lateral sclerosis (*ALS*)
- Infantile progressive spinal muscular atrophy (*Type 1, Werdnig-Hoffmann disease*)
- Intermediate spinal muscular atrophy (*Type 2*)
- Juvenile spinal muscular atrophy (*Type 3, Kugelberg-Welander disease*)
- Adult spinal muscular atrophy (*Type 4*)
- Spinal-bulbar muscular atrophy (*Kennedy disease*)

**Inflammatory Myopathies**
- Polymyositis
- Dermatomyositis
- Inclusion-body myositis

**Diseases of Neuromuscular Junction**
- Myasthenia gravis
- Lambert-Eaton (myasthenic) syndrome
- Congenital myasthenic syndromes

**Diseases of Peripheral Nerve**
- Charcot-Marie-Tooth disease
- Friedreich’s ataxia
- Dejerine-Sottas disease

**Metabolic Diseases of Muscle**
- Phosphorylase deficiency (*McArdle disease*)
- Acid maltase deficiency (*Pompe disease*)
- Phosphofructokinase deficiency (*Tarui disease*)
- Debrancher enzyme deficiency (*Cori or Forbes disease*)
- Mitochondrial myopathy
- Carnitine deficiency
- Carnitine palmityl transferase deficiency
- Phosphoglycerate kinase deficiency
- Phosphoglycerate mutase deficiency
- Lactate dehydrogenase deficiency
- Myoadenylate deaminase deficiency

**Myopathies Due to Endocrine Abnormalities**
- Hyperthyroid myopathy
- Hypothyroid myopathy

**Other Myopathies**
- Myotonia congenita
- Paramyotonia congenita
- Central core disease
- Nemma myopathy
- Myotubular myopathy
- Periodic paralysis

MDA’s website, mda.org, is constantly updated with the latest research news and information about the diseases in its program. Follow MDA on Facebook, Twitter and YouTube.

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