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The CMTA Report

APRIL 2001

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A resource for information on Charcot-Marie-Tooth disease (Peroneal Muscular Atrophy or Hereditary Motor Sensory Neuropathy), the most common inherited neuropathy.

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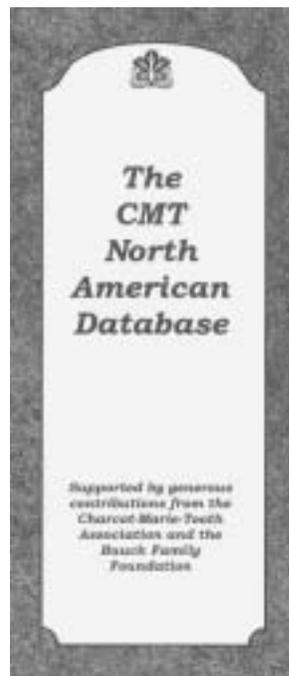
The CMT North American Database: A Giant Step Forward on the Road to A Cure for CMT

By **ANN LEE BEYER, Chairman**
CMTA Board of Directors

The CMT North American Database is up and running. We are getting phone calls and the packets are ready and waiting to mail or download. Dr. Shy reports that he has already received a number of completed forms. This is a very exciting project and will make an enormous difference in the lives of all of us who are affected by CMT.

The CMTA is in its eighteenth year of serving the CMT community and we are thoroughly committed to living our Mission "to improve the quality of life for people with CMT." In honoring this commitment, the CMTA does more to advance the cause of CMT than any other organization. We do this by providing education and support through our literature, in particular, our bimonthly newsletter, *The CMTA Report*; the genetics guide for patients; the CMT website; and CMTA patient/family conferences; as well as through our national chain of CMT support groups. In addition, we staff booths at medical conferences so that clinicians and researchers can learn about CMT and the CMTA. We are very fortunate to have an active, involved Medical Advisory Board who work with us to encourage and support

Please call the CMTA for your database packet today or download the survey from our website at www.charcot-marie-tooth.org. Your participation will help make the database a valuable research tool.



CMT research and sponsor international research symposia.

Although the above-mentioned programs are important, our ultimate goal is a cure. We are making strides. The CMT North American Database is probably the most important project we have ever undertaken towards attaining this goal.

HOW THE DATABASE WILL HELP PATIENTS

First, the information gleaned in the database will give physicians and researchers a more thorough understanding of all the clinical symptoms that accompany the various types and subtypes of CMT, thus allowing them to say with certainty, "This is what CMT actually is."

(continued on page 2)

The CMTA.... Living our Mission

Mission Statement:
To improve the
quality of life for
people with CMT.

Vision Statement:
Dedicated to
generating hope.

CMT DATABASE

(Continued from page 1)

Second, it will provide clinicians and researchers with a better picture of the severity and progression of this disease.

In addition this knowledge will also:

- Make it easier to diagnose.
- Be a blessing for those who have lived—sometimes for years—with the uncertainty of whether or not they have CMT.
- Provide a better picture of how prevalent CMT actually is.
- Influence and probably change some of the directions that CMT research will take.
- Stimulate research interest and attract more research funds.
- Speed up the pace of CMT research by making the information gleaned in this database available to CMT researchers.

As was pointed out in our last issue of *The CMTA Report*, “rapid advances in molecular genetics have generated great excitement in the field of CMT disorders.” The more people enrolled in the database, the more information will be available to researchers. In order to gather enough information to make a difference, we need as many people as possible to enroll in the

*The CMTA
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CMT Research Campaign Begins

By the time you receive this newsletter, you should have also received a request to help fund our ambitious research goals for next year. Among our projects is the groundbreaking North American Database, about which you have just read. We will also continue to award grants to individual researchers who seek to unravel the mysteries of the gene defects which cause CMT’s various subtypes and to fund studies which seek to improve the lives of people currently living with CMT.

Be a contributor by completing the survey forms which will help give substance to the database and by financially supporting our grants and fellowships!

The CMTA seeks to help its members and asks the help of its members in supporting its ambitious goals.

database. If you don’t volunteer, how are we ever going to learn about CMT? Remember, everyone who enrolls will be assigned a number to protect his or her identity. There are many safeguards in place to ensure this.

HOW TO GET A PACKET

The CMT Database packet can be obtained by calling the CMTA office (800-606-CMTA), or getting in touch with Wayne State University in Detroit (313-577-5273). It can also be downloaded from several websites:

- The CMTA website (www.charcot-marie-tooth.org)
- Wayne State University (www.med.wayne.edu/neurology/cmt.html)
- The Neuropathy Association (www.neuropathy.org)
- The MDA (www.mdaua.org)

In addition, it will soon be on the American Academy of Neurology’s website (www.aan.com).

Information about this project also appears in the American Neurological Association’s latest newsletter, and in the most recent issue of the *Quest* magazine.

THE SOURCE PERSON

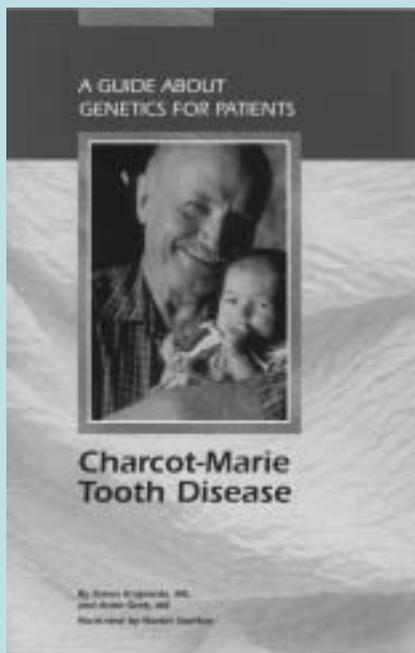
When you receive your Database packets, you will notice that it is suggested that one person—who must have CMT—become the Source Person for your family. However, if other family members are completing questionnaires, there may be more than one Source Person.

For example, you may be the Source Person for your immediate family but have a daughter, son, sister, first cousin, or second cousin who also has CMT in her or his family, and s/he will be contacting the CMTA or one of the above providers, to request the Database packet. Whoever fills out the forms in that branch of the family will list her or himself as the Source Person for that branch. Not to worry! The design has been well thought out and it all makes sense.

If you have any questions about participating, Ravin Lewis at Wayne State University (313-577-5273) is available to help you.

Please help us help you. Call the CMTA to order the CMT North American Database packet. Fill it out. Return it in the self-addressed envelope. Or go onto one of the above-listed websites and download the packet.

Together we will make a difference!



GENETICS BOOKLET Now Available

The long-awaited booklet on the genetics of CMT is available from the CMTA for the price of \$4 for current members and \$5 for non-members. All proceeds from the sale of this booklet will go to fund research to help in the ultimate cure of CMT.

Not only does the booklet explain, in detail, about CMT1A, 1B, CMTX, and HNPP, but it also explains autosomal recessive inheritance patterns. Because the type 2 variations of CMT have not yet been linked to the causing genes or the specific mutations, those forms are not discussed in detail.

Some commonly asked questions, such as "If someone in my family does not have symptoms of CMT, can he or she have a child with CMT?" or "Why am I the only one in my family with CMT?" are covered in the closing pages of the booklet.

In a compact and easily read format, this little booklet, "A Guide About Genetics for Patients: Charcot-Marie-Tooth Disease," is a must-have for every patient library.

CMTA MEMBERSHIP/ORDER FORM

Name: _____

Address: _____

Phone Number: _____ Email: _____

Members who are current with their dues are considered "active."
If you are unsure as to whether you are current with your member dues, please call the office at 1-800-606-CMTA.

	QTY	COST	TOTAL
Charcot-Marie-Tooth Disorders: A Handbook for Primary Care Physicians		active members \$15 inactive members \$20	
Membership Dues		\$35	
SPECIAL OFFER FOR ACTIVE MEMBERS ONLY: CMT Facts Series (I-IV)		active members \$16	
CMT Facts I <input type="checkbox"/> English <input type="checkbox"/> Spanish		active members \$3 inactive members \$5	
CMT Facts II <input type="checkbox"/> English <input type="checkbox"/> Spanish		active members \$5 inactive members \$7	
CMT Facts III		active members \$5 inactive members \$7	
CMT Facts IV		active members \$8 inactive members \$10	
A Guide About Genetics for the CMT Patient <i>NEW!</i> <i>No shipping and handling on this item only.</i>		active members \$4 inactive members \$5	
VCR Double Tape: duPont Conference		active members \$25 inactive members \$40	
CMT Informational Brochure <input type="checkbox"/> English <input type="checkbox"/> Spanish		FREE	
Physician Referral List: States: _____		FREE	
Letter to Medical Professional with Drug List		FREE	
Contribution to CMT Research <i>10% will be applied to administrative expenses.</i>			
Shipping & Handling <i>Orders under \$10 add \$1.50, orders over \$10 add \$3.00</i>			
TOTAL			

Check payable to the CMTA (US Residents only).
Foreign residents, please use a credit card or International Money Order.

VISA MasterCard American Express

Card Number _____ Expiration Date _____

Signature _____

Mail to the CMTA, 2700 Chestnut Parkway, Chester, PA 19013

A copy of the official registration and financial information may be obtained from the Pennsylvania Department of State by calling, toll-free, within Pennsylvania, 1-800-732-0999. Registration does not imply endorsement.

Gait Analysis: A State-of-the-Art Diagnostic Tool

By **ADRIAN B. LIGGINS, PhD, PEng**, Director, Motion Analysis Laboratory,
Shriners Hospital for Children, Philadelphia, PA



VISIT OUR HOMEPAGE

at

www.charcot-marie-tooth.org

For a patient with a neuromuscular disorder, a typical clinic visit may include walking while being observed by a clinician. As the patient walks, the clinician uses visual clues to put together a picture of his or her gait (or, walking pattern). This information, along with other clinical tests (such as range-of-motion tests or muscle-strength tests), is used to determine the possible causes of any gait anomalies and the potential treatment options. However, this approach places undue reliance on our (somewhat limited) ability to visually observe and interpret the complex motions which make up walking.

As far back as the late 1800s, photographic techniques were used to freeze motion, allowing it to be examined in greater detail. The most famous example is Edward Muybridge's work showing that a running horse momentarily had all four hooves off the ground in unison. Muybridge went on to examine human gait and other daily activities. During the next century, improved techniques were developed for recording and examining walking patterns; however, until the advent of suitably powerful computers in the latter part of the 20th Century, the analysis of human gait was too cumbersome and labor-intensive to be applicable in a clinical setting.

The modern Gait Analysis Laboratory (also known as a Motion Analysis Laboratory) uses a collection of state-of-the-art techniques and equipment to comprehensively assess a patient's gait and the underlying reasons for that gait. These techniques may include:

Clinical Examination: A physical therapist assesses how far each joint (hip, knee, and ankle) will move (its "range of motion") and how much force the muscles can create around each joint. This information gives a clearer understanding of the patient's physical limitations to walking.

Video Analysis: The patient wears a t-shirt and shorts and walks up and down a walkway marked on the floor. Video cameras record the patient from the front and side. This procedure provides both an overall view of the patient walking, and rudimentary information, such as

Gait analysis gives a clearer understanding of a patient's physical limitations to walking.

whether he walks on his toes, is crouched (bent at the hip, knee, and ankle), or uses a lot of trunk motion during walking.

Motion Analysis: Special reflective markers are placed on the skin, using double-sided tape. These markers (usually thirteen or so in total) are positioned over points on the pelvis, femur (thigh bone), tibia (calf bone), and foot where an easily located bony prominence lies just under the skin surface. As the patient walks down the walkway, six special cameras, spaced around the laboratory, send images of the reflective markers back to a central computer. From these images, the computer reconstructs the positions of the markers, allowing bone positions to be determined. The motions of the pelvis, hip, knee, and ankle can then be mapped out, and compared to those for a typically developing subject, allowing the type and magnitude of any differences to be identified.

Muscle Activity Analysis: Electrodes (small plastic devices with four small metal pads) are placed on the skin, over the major muscle groups used for walking. These devices pick up the electrical signal from the muscles as they switch on and off during walking, and relay this information to the central computer. From here, inappropriate muscle activity, which may lead to walking problems, can be identified.

Muscle Force Analysis: As the patient walks down the walkway, panels in the floor record the forces between the foot and the floor. This information allows the forces and powers generated around the hip, knee, and ankle to be calculated. These forces and powers are compared to those for a typically developing indi-

ool for Walking Problems

vidual: differences may indicate weaknesses, or potentially detrimental compensations (for instance, excessively high forces might be generated around a bent knee, to prevent it bending further).

Foot Pressure Analysis: The patient walks over a special panel in the floor, which measures the pressures across the undersurface of the foot. This procedure can indicate unusual pressure-distributions and show how a walking problem is altering the way in which loading is transmitted between the foot and the floor.

With all this information being collected, a patient visit may last up to three hours. The information is then analyzed, interpreted, and compiled into a report for the patient's referring physician. This report, plus all the information detailed above, may also be reviewed at a clinical meeting. The end-product is a list of recommendations and options for treatment: such treatment may be therapy, bracing, or surgery. If surgery is performed, a gait analysis is conducted one year post-op to assess the patient's outcome and progress.

The main benefit of gait analysis is the depth of information that can be gathered in order to understand a patient's walking pattern and the underlying biomechanics of any gait anomalies. Clearly, this information is far beyond that which can be gathered with the naked eye. In addition, a greater number of related professionals are involved in the decision-making process: gait laboratories are, by definition, multi-disciplinary (our laboratory includes an engineer, a biomechanist, a kinesiologist, and physical therapists), plus a clinical review meeting will include an attending orthopaedic surgeon and additional clinical staff. As such, up to ten professionals may ultimately be involved in the decision-making process.

For all the advantages that they have to offer, gait analysis laboratories are still relatively uncommon: it is estimated that there are only 120 or so clinical labs in the United States. Fee-for-service costs may also be high (often over \$2,000 per patient assessment). Shriners Hospitals for Children currently owns twelve Motion

Analysis Laboratories: a further lab is currently under construction and there is a long-term desire to provide each of the orthopedic hospitals within the Shriners system (19 of the current 22 hospitals) with this facility. To our knowledge, this is the largest group of gait analysis labs under a single organizational umbrella in the world. As with all care in Shriners Hospitals, gait analysis is free-of-charge (paid for by the Shriners organization). Our laboratory assesses children from five to eighteen years of age, on referral from their Shriners physician.

For more information, visit these websites:

- **Shriners Hospitals:**
www.shrinershq.org/Hospitals
- **Clinical Gait Analysis:**
<http://aisr1.lib.tju.edu/cga>



A patient demonstrates the computerized aspect of motion analysis at the Shriners Hospital in Philadelphia.

Why Does Peripheral Neuropathy

By **STEVEN SCHERER, MD, PhD, William N. Kelley Associate Professor of Neurology, The University of Pennsylvania Medical Center, Philadelphia, PA**

(Editor's note: This information was presented as part of Dr. Scherer's presentation at the CMTA patient/family conference at the University of Pennsylvania Medical School.)

Peripheral nerves are a collection of nerve fibers that originate from many different kinds of neurons. Motor fibers originate from motor neurons that are located in the spinal cord. Sensory axons originate from neurons that are located outside the spinal cord in large clusters called ganglia. The ganglia that contain the sensory neurons for the leg are located in the low back region (called the lumbar and sacral levels); those for the arm are located in the neck (called the cervical region). Each of these ganglia contains many thousands of sensory neurons.

Every sensory neuron has two ends. One end is connected to a tissue in the body (a piece of skin, muscle, bone, etc.), and the other end is connected to the spinal cord. Under normal circumstances, sensations are generated only upon stimulation of the end of the nerve fiber that is in the body. Then sensory nerve fibers relay this information to the spinal cord, and cells in the spinal cord, in turn, relay this information to the brain.

There are many kinds of sensory neurons. This is why we can perceive so many different sensations. All of us can appreciate many of these sensations, such as heat, cold, light touch, pin prick, vibration, and movements of the hairs on our skin. Other sensations are less obvious, such as our ability to determine movements of our arms and legs. Each kind of sensation, including pain, is conveyed to the spinal cord by certain kinds of sensory neurons.

So what does this have to do with pain? It is likely that some kinds of neuropathy damage the sensory fibers that convey pain, causing them to be hyperactive even in the absence of stimulation. In other words, damaged "hyperactive pain fibers" trick the brain into perceiving a painful stimulus even though none is present.

The hyperactive fibers may not even be properly connected to their tissue, thereby accounting for why people can experience pain in their numb feet or legs.

It should be clear that not all pain is caused by neuropathy, even in people who have peripheral neuropathy. The pain of arthritis and headache, for example, is conveyed, but not caused, by sensory fibers. Even the pain caused by one of the foot deformities caused by neuropathies is not caused by damaged sensory fibers; the sensory fibers are merely conveying the information to the spinal cord. Conversely, not all people who have peripheral neuropathy have painful symptoms. Pain is a common symptom in some kinds of neuropathy, such as diabetic neuropathy, in which small sensory fibers may be disproportionately affected. Among people who have inherited neuropathy, pain is much less frequent in the demyelinating forms than in the axonal forms affecting small sensory fibers.



Dr. Steven Scherer, a recent addition to the Medical Advisory Board, believes pain management should "maximize a patient's quality of life."

THE PRINCIPLES OF TREATING PAINFUL PERIPHERAL NEUROPATHIES

If neuropathy causes pain that is diminishing the quality of life, then this symptom should be treated. In my view, to manage pain effectively, there has to be a partnership between the patient and the physician. The patient needs to understand the pain—when it occurs, how well the drugs work, the side effects of the medications (particularly how troubling they are)—and communicate these things to the physician. The physician needs to know the medications and the relevant information about them—their duration, common side effects, potential interactions with other drugs, and whether a patient has other complicating medical problems—and communicate these things to the patient.

The goal is to maximize the patient's quality of life. In practical terms, the patient should take the amount of medication that effectively manages the pain, but that does not cause unaccept-

Cause Pain?

able side effects. In the ideal case, the patient would be pain-free without any side effects. In the worst case, the patient has intolerable side effects at a dose that produces no pain relief whatsoever. In the typical case, however, there is a dose of a medication that provides some pain relief but that also causes some side effects. It should be clear that only the patient can know whether a medication works and whether it has acceptable side effects.

MEDICATIONS FOR TREATING PAINFUL PERIPHERAL NEUROPATHIES

Many medications have been reported to work for painful peripheral neuropathies. A few have been studied in rigorously conducted clinical trials, such as desipramine for painful diabetic neuropathy. Several more have been reported to be effective for painful neuropathy, including other kinds of pain syndromes such as post-herpetic neuralgia or trigeminal neuralgia. I am not aware of any studies that have specifically examined treating pain in inherited neuropathies. Regardless of the medication, the logic is the same:

- Introduce one medication at a time. Changing the doses of two medications simultaneously makes it difficult to determine which medication is responsible for any given effect (especially a side effect).
- Use a gradually escalating dose of one medication until either good pain relief is obtained or intolerable side effects occur. This is the key concept; too often I have seen patients who have been taking potentially effective medications but at doses that neither help the pain nor cause significant side effects.
- If one medication fails, try another one.

The medications that work for the pain of neuropathy fall into a few groups:

Tricyclics (amitriptyline/elavil, nortriptyline, desipramine/norpramin). These drugs were originally used as antidepressants, typically at much higher doses than are used for treating painful neuropathies. They probably work by blocking norepinephrine receptors. They are

usually taken once a day, an hour or so before sleep, as they are slowly metabolized, and often cause some degree of drowsiness or sedation. The drowsiness is often a useful side effect when pain interferes with sleep. One typically starts with a low dose (25 mg or even 10 mg) and “builds up” the dose until either a good effect has been achieved or there are intolerable side effects (typically 50–100 mg). Beside drowsiness, a dry mouth and cognitive side effects are common (and there are other side effects, too). It is important to know that the tricyclics typically take 2–4 weeks to reach their full effectiveness against pain, and that the severity of the side effects often diminishes over time.

Neurontin (gabapentin). This medication is not approved for the treatment of chronic pain, but is probably more widely used for pain than for the treatment of its approved indication, epilepsy. It was designed to be a long-lasting mimic of a neurotransmitter, GABA. Neurontin comes in 100-, 200-, and 300- mg capsules; these are taken every 6–8 hours (the dose to be determined by its efficacy and side effects!). Cognitive changes are the most common side effect. In my experience, Neurontin works less reliably than do the tricyclics.

Narcotics. The keys for using narcotics are matching the duration of action to the duration of pain, and letting the patient figure out the dose that provides adequate pain relief with acceptable side effects. There are a few kinds of long-acting narcotics:

- MS Contin (the active ingredient is morphine; 15-, 30-, 60-, 100-mg tablets); works for about 12 hours.
- Oxycontin (the active ingredient is oxycodone; 10-, 20-, 40-mg tablets); works for about 12 hours.
- Duragesic patches (the active ingredient is fentanyl; comes in 10cm²/2.5 mg, 20cm²/5.0 mg, 30cm²/7.5 mg, and 40cm²/10 mg size patches); works for 2–3 days/patch.

There are many kinds of short-acting narcotics (I often use the regular form of

(continued on page 15)

HNPP Is Not Always Painless

By MAUREEN HORTON, RN

Hereditary neuropathy with liability to pressure palsies (HNPP) is typically described as painless, recurrent episodes of pressure palsies caused by pressure, stretch, or repetitive use. This is the classic form of HNPP, only one of the five main phenotypes of the disease. Little, if anything, is written about pain in HNPP.

Once a person is finally diagnosed with HNPP, he or she hears from his or her doctor that it is a “pain-free, no-big-deal-disease,” or, as one individual was told, quite emphatically, “HNPP does not cause pain!” This is great news for the approximately 90% of individuals who do not have pain, but for the estimated 10-15% who do have pain, it can be quite frustrating to hear.

As one woman wrote, “At my last visit to the neurologist, he told me that HNPP was painless, that it was just a numbing problem. When my husband and I left, I just

wanted to cry. I told my husband that I knew I had HNPP from the DNA, but there must be something wrong with me elsewhere—cancer or something—because of the pain. I knew I wasn’t crazy, but I felt like the doctor thought I was...”

As I wrote, with the help of Drs. Garcia and Parry, in the February/March 1999 edition of *The CMTA Report*, describing both CMT and HNPP, “Pain may occur...but it is usually not “neuropathic” pain. Cramps are frequent, most often in the limbs (legs and arms) and are produced by overuse of weak muscles. Low back pain is frequent and is due to the strain produced by the weak leg muscles and overuse of the paraspinal and back muscles. Back pain is frequently relieved by AFOs. Although it is not common, “neuropathic” pain definitely does occur in a portion of patients. It consists of aching, burning and shooting pain in the distribution of affected nerves.”

The HNPP e-mail support group is skewed toward individuals who have more involved or severe cases of HNPP. Those who find this disease to be more than a nuisance are looking for answers and support. Most have signs of the beginning of a more generalized neuropathy.

They also have a higher percentage of pain. Yet, few, if any, report pain during what they consider to be a pressure palsy episode.

They do have cramping pain in their toes, feet, legs, fingers, hands, and even their necks. A few have mentioned “charley horse” type pain in their abdomens, set off by bending over. Despite attempts to stretch them out, some cramps can last up to a few hours. Some patients have muscle spasms in their backs related to activity.

Aching pain is mentioned by many. Again, the pain involves hands, arms, feet, ankles, legs, and oh, those aching backs! Some patients attribute it to some activity around the house, or too much activity or just walking “funny” or exercise. Some think it is due to weak muscles. Another pain problem is related to not moving. Most people within the group can identify with the uncomfortable sensation we often get that tells us it is time to reposition or we will go numb. Yet some people also have actual pain if they have been in one position too long. A

few are even awakened from sleep by sharp pain, which disappears once they move around.

“Zingers,” as my dad calls those very sharp shooting, electric-like pains, are not uncommon. They run along the path of a nerve and typically last a few seconds at most. Yet some people have repeated episodes of these, over a few minutes. Some individuals have these rarely, but for others it is a daily or hourly occurrence. This is a form of “neuropathic” pain. Neuropathic pain can also take the form of deep aching or burning or sharp pain running along the nerve.

Some people can tell when a low front is coming through or the barometer is dropping. A few women have noticed more problems during pregnancy or surrounding their menstrual periods. Two individuals in the group report mouth and throat burning, while another two have headaches. Whether mouth burning and headaches have anything to do with HNPP is not known yet.

Other people with HNPP develop muscle sprains or strains from twisted weak ankles or overuse. These are treated in the usual manner of ice, heat, rest, and splinting, if needed.

How pain is treated is as varied as the pain itself. It is dependent on how often and severe the

For the estimated 10-15% who do have pain, it can be quite frustrating to hear that HNPP is a “pain-free, no-big-deal-disease.”

pain is, how much one is bothered by it, how one wants to treat it, and the doctors one has on their medical team. When pain is infrequent or mild, treatment may not be warranted. My dad, for instance, is really bothered by “zingers,” but they only happen a few times a week and last a split second. While drugs could potentially help, he does not have problems often enough for him to want to take drugs. Others try over-the-counter medications, vitamins, splints, showers, massage, changing positions, stretching, rubbing, rest, pacing activities, and anything else that works to help with the pain.

When the above methods no longer work or the pain is worsening or interferes with activity during the day and sleep at night, more help is called for. Pain is very fatiguing and the fatigue of pain can be another reason to seek treatment. But finding the right help can be problematic. One individual wrote, “I asked my doctor about the pain in my hands and legs and why it is getting worse, and he said “well, there is nothing I can do about it. Just live with it!” You don’t have to “just live with it.” There are medications which can do amazing things to help HNPP pain. Some of the antidepressant medications, such as Elavil, can help nerve pain. This is not to say that one is depressed, or that the pain is in one’s head. These medications at lower doses than used for depression do work on neuropathic pain. Some of the anti-seizure drugs, such as Tegretol and Neurontin, work amazingly well also. These two classes of drugs are the main arsenal in the treatment of neuropathic pain. It may take one class of drug or a combination of them. And it will take time to find the right dosage.

Not all neurologists are comfortable treating pain. If your neurologist seems to be uncomfortable treating you, a referral to a doctor or clinic that specializes in pain may be a better option. I have found there seem to be two different pain clinic philosophies. One seeks to get the patient off the medications (especially any narcotics) and uses behavior modification to treat pain. The second philosophy seems to be more willing to use narcotics as well as the behavior modification as needed. Besides finding a physician or clinic that matches your philosophy of dealing with pain, it is imperative that the doctor be interested in learning at least the basics of HNPP.

As Dr. Parry wrote in *The CMTA Report* (Vol. 17, No. 2, April/May 2000), “The effects of the disease on each patient are as unique as the individuals themselves.” This is as true of their pain problems, and how they treat them, as it is of all their other symptoms. Pain is treatable. You don’t have to “just live with it!”

Medical Information on the Internet

Finding information on the Internet is becoming easier and easier as technology improves, but finding accurate information, especially as it relates to health issues remains a problem. Anyone can produce a website or web page, but a consumer needs to be able to tell if a site contains reliable medical information.



A reliable site will clearly disclose what group or person maintains the site and will display a phone or e-mail contact along with a statement of the site’s goals. All health-related information should be clearly dated and references or links to scientific literature should be included. In general, health sites maintained by the federal government, universities, research centers, and respected national associations are likely to contain the most accurate information.

Some of the best places to find information related to Charcot-Marie-Tooth disorders are:

- **The Charcot-Marie-Tooth Association** home page at www.charcot-marie-tooth.org. Remember that discussion forums and chat rooms are places for lay persons to talk about their own problems and successes and are not locations for reliable, verifiable medical information.
- **The Muscular Dystrophy Association** can be found at www.mdausa.org. This site has an extensive section called “Ask the Experts” in which questions and answers specific to CMT can be found. The current and back issues of *Quest* magazine are also found here.
- **The Neuromuscular Diseases Clearinghouse** is maintained by the University of California, Davis, and can be found at www.medpmr.ucdavis.edu. This site contains a section called “Tools for Daily Living,” which has links to tips on alternative medicine, disability rights, and traveling with a neuromuscular disease.
- **The National Institutes of Health** has a searchable database at www.clinicaltrials.gov which provides centralized information on clinical trials funded by the NIH.
- **The National Library of Medicine**, supported by the NIH, has a free, searchable database of medical-related scientific literature at www.ncbi.nlm.nih.gov. If you type in Charcot-Marie-Tooth, you will see a list of journal articles that include those key words. You can double click on any article title and see a brief abstract. This is a very technical site, used primarily by doctors and researchers.
- **The Washington University School of Medicine** maintains the Neuromuscular Disease Center site, which is found at www.neuro.wustl.edu/neuromuscular. This site is somewhat technical and is designed for scientific and medical personnel, but it does contain details about disorders and their subtypes, including symptoms, genetic defects, rate of progression, etc.

First Steps to Freedom

By KATE MCKENNA (Excerpted from *The Washington Post*, with permission)

It was March 1999 in Pristina, Kosovo's capital. Masked Serbian militia marauded through the city, ordering all ethnic Albanians out of their homes immediately. As frightened residents crowded into the streets, the night sky glowed from the torching of distant houses. Shots rang out and the entire Ademi family—mother, father and four children aged 14 to 22—knew there was no time to lose.

But the Ademi's situation was unlike that of their fellow Kosovo Albanians. Adding to their terror and vulnerability was the fact that three of the four children were crippled by a mysterious condition that impeded simple mobility—much less a forced trek to the border at gunpoint.

The oldest son, 22-year-old Artan, could no longer walk unassisted on his twisted limbs; 14-year-old Valon would lose his balance and fall every few steps. And Fllanza, 17, was bedridden, her ankles turned sharply inward and frozen at unnatural angles. In valiant efforts to stand, she had broken her feet and raised calluses on her ankles. She was now reduced to crawling painfully on her hands and swollen knees. Doctors in Kosovo had told the family that nothing could be done, that Fllanza and her brothers must simply accept their fate, as this unexplained malady slowly curled their limbs into bent and useless things.

For most of her teenage years, Fllanza was forced to crawl on her hands and knees. Now, following surgery in the United States, she can walk again.



But now, they had to move or die... “The soldiers didn’t care if you were sick, if you could walk or not,” she (the mother, Fehmije Ademi) says. “They were barbarians. They would grab you, and push you, and kick you. And the ones who couldn’t move fast enough, they would shoot.”

...But, what happened to this family in the following months was little less than a miracle. The tragic circumstances of their upheaval notwithstanding, this episode of “ethnic cleansing” led them, oddly enough, to salvation.

At a resettlement camp, the unexpected intervention of an American delegation gave them a chance at a new life and medical attention. From the burning ruins of their life in Pristina, they came to a place where their condition was seen not as the unalterable hand of fate, but as a recognizable disease that could be treated, if not cured—a disease with a name, albeit an odd one: Charcot-Marie-Tooth.

...Artan Ademi was just past puberty when the disease became evident, bending his feet inward at freakish angles and impeding his ability to move. Medical experts call this symptom “marionette gait” because it leaves the legs and hips unstable and makes walking an arduous affair. But at least he was still able to put weight on his feet.

Fllanza was less fortunate. At about age 13, her feet contorted inward, then froze in that position. Her parents, who had watched the disease cripple their oldest son, were inconsolable. “We lived the sadness twice,” recalls Fehmije, speaking through an interpreter. Fllanza tried to keep walking—on the outer edges of her twisted ankles—but for most of her teenage years, crawling on her hands and feet was her only means of locomotion. The family could not afford a wheelchair.

Then, the Ademis’ youngest boy, Valon, developed symptoms. His deformities worsened until he was able to walk only on his toes.

Kosovo doctors offered no consolation, appearing mystified by the family’s misfortune. “They said it was God’s will,” says the children’s father, Nazmi Ademi, in a voice filled with sadness. “It’s a disease, and there’s nothing we can do,” they told him.

Miracles abound in this story... Family members say their first miracle was finding each other again, unharmed, at a Macedonian refugee camp after weeks of separation and deprivation... Lost in the turmoil and forced to hide in

the hills for weeks, Artan and Fllanza were the last to reach the relative safety of the camp... Fellow refugees slowed their own pace to hoist Artan upright; others dragged Fllanza along for miles in a blanket.

Once reunited, the family drew notice from relief workers. "Their condition was a big shock to everybody there," said Ingrid Bregasi, the family's interpreter. "It helped them get the attention they needed."

Lutheran Social Services of the National Capital Area got a call from abroad, asking if the agency was willing to take on a family with so many needs. "Medical cases need a lot more time and attention than the typical refugee/asylum case. And we had no idea what their condition was," recalls Ruth Anne Dawson, director of the agency's Falls Church, VA, office. "We only knew they were in wheelchairs (that had been provided by relief agencies). We knew (the disease they shared) was genetic. But we didn't know anything else about it." Possible diagnoses ranged from advanced arthritis to muscular dystrophy to polio.

In August 1999, the Ademi family was settled in a first-floor apartment in Alexandria. Over the next six months, the Ademis went from doctor to doctor—internists, neurologists and foot doctors... But doctors repeatedly declined to take their case, dashing their new hopes.

Richard Foa, then a neurologist at Georgetown University Hospital, was the first to diagnose Charcot-Marie-Tooth that fall... Foa recalls that the Ademis presented a unique and extreme case. "It's highly unusual to see three out of four siblings heavily affected and one spared totally. And for it to be so forcefully manifested in the children, but not in the parents, that also is unusual." Also startling, "particularly to Western eyes," he says, was to see a condition that had gone neglected so long.

...The family eventually came to Paul S. Cooper, director of the Foot and Ankle Center at Georgetown. It was Cooper who literally put the Ademis back on their feet again. Taking on their case *pro bono*, he made plans for immediate treatment, including surgery.

Treating such extreme cases of the disease was a first for Cooper, despite his years of experience working with CMT patients at Georgetown and in Connecticut. He chose a series of surgeries, involving slicing into bones and transplanting muscle from an unaffected part of the leg to an impaired part. "Nothing we did was uniquely revolutionary separately—but the combination of all these treatments performed on one patient is fairly unique," he said.



...First, Cooper rebuilt Fllanza's foot by fusing three bones (triple arthrodesis) below the ankle to provide greater strength and allow Fllanza maximum motion in that joint. Next, he transferred tendons from the strong side of her foot to the weak side, in a soft-tissue balance procedure often used for stroke and polio patients. The goal was to correct the imbalance of foot muscle strength that pulls the foot in unusual directions.

Those same two procedures would put Artan, the oldest son, on the road to recovery. One similar, less radical, operation took care of Valon's toe deformities and tendon problems. But Fllanza's condition was so severe that it required another delicate procedure.

...So as not to risk further damage to nerves and arteries, he installed an Ilizarov frame, consisting of high-tension wires cutting through the skin to the bone. The device is generally used to save limbs of diabetics and patients with infections who might otherwise face amputation.

Through a series of color-coded struts, the device allowed Fllanza's feet to be moved slowly back into position, millimeter by millimeter. Her mother tightened the settings daily to keep the strings taut, like those of a well-tuned piano.

The day Fllanza took her first halting steps—four months after the operation and nearly five years since she'd last walked normally—her mother cried all day. "It was such a miracle," she says now. "I had hopes, but never could have imagined how good it would be."

(continued on page 12)

Fllanza happily does her physical therapy and strengthening exercises, hoping one day to dance.

FREEDOM

(Continued from page 11)

Today, Fllanza wears a constant smile. "She was even happy to get into the surgery room," says Bregasi (their interpreter). "Now she's so happy, she smiles all the time!" Even the aftermath of surgery, the discomfort of the Ilizarov frame, the wires through her skin and painful physical therapy didn't take the grin off her face.

Once again, she's walking—haltingly, but without cane or walker. While doctors will always need to monitor her condition, particularly her hips, knees and hands, for signs of muscle wasting or weakness from the progressive disease, therapy has stabilized her feet. Now she says she knows she will one day live on her own, hold a job, drive a car. A once-bleak future suddenly holds all kinds of possibilities.

Artan and Valon, say their parents, are similarly excited... "Imagine if they were still in their country," Bregasi says. "Maybe Fllanza would be

crippled for the rest of her life. And her brothers, the same thing. A lot of good happened that may never have happened if they had not been told to leave."

There is much to hope for. Fllanza, at 19, hopes to be able, once again, to dance the "shotave"—a traditional Albanian dance—and recover a portion of the youth she lost in Kosovo. Artan and Valon want to learn to drive. They agree their journey has been miraculous in many ways, but find it hard to describe, in any language, the dramatic changes the last two years have brought to their lives.

"There are no words to describe what has happened, and what these doctors have done for them," says their father, shaking his head slowly and looking at his now-vital daughter and sons. "There are no words."

Editor's Note: A side bar that accompanied the article listed the CMTA with its 800 number as the source of additional information on CMT and has resulted in many calls to the office.

Individuals Making a Difference

Golf Tournaments Raise Funds for CMT Research

Two New York Area CMTA golf outings last year were each a huge success, raising approximately \$41,000 for CMT research. All of the elements required to assure the success of fundraisers were in place: a generous underwriting sponsor, an elite golf course, excellent food, super prizes, and the total support of organization members in planning the event and providing monetary support.

At this time, the CMTA Board has been working with other individuals to organize golf outings to raise funds for CMT research. In 2001, at least three golf tournaments are scheduled. In each case, one individual or family has taken on the responsibility of organizing and running the event because they believe so strongly in the work that is being done to advance the cause of CMT research.

Richard Sharpe and Robert Kleinman are, again, working on a New York golf out-

ing in the fall and Patrick Torchia has organized a similar tournament in Johnstown, PA.

The Wayne State University CMT Clinic is hosting its first annual golf tournament on Monday July 23, 2001. The tournament will be held in Ann Arbor, Michigan at the Barton Hills Country Club, one of the finest courses in Michigan. All proceeds from the event will be used to support patient care and research at the clinic. Those interested in participating should contact Lisa Stelter at lisa@lisastelter.com.

These tournaments demonstrate how much can be done by one or two individuals in supporting the cause of research.

Richard Sharpe (Treasurer of the CMTA) (516-656-0681) would be willing to share his experience and expertise in running a golf outing with any other members who might want to be part of such a fundraising opportunity.



Professionals Working for CMT Patients: Dr. Carlos Garcia

(Editor's note: Dr. Garcia has been a valued member of the Medical Advisory Board for years and will be hosting the New Orleans patient-family conference in April at Tulane University School of Medicine. He is a naturalized American citizen, and is married and the father of three girls.)

Dr. Garcia was born in Cali, Colombia, South America and was educated there, receiving his MD from the Universidad del Valle. He came to New Orleans in 1962 as a Kellogg Foundation fellow in Neuropathology to work with Dr. John Moossy. He did his neurology residency in adult neurology at the Charity Hospital of New Orleans from 1965–1967, during which time he was an NIH fellow. He is currently Board certified in anatomic pathology, neuropathology, adult neurology and neurorehabilitation.

Dr. Garcia was the clinic director for the Muscular Dystrophy clinics in south Louisiana for over 20 years. He is now the MDA clinic director in Lafayette, LA and runs a neuromuscular clinic and the neuromuscular histopathology laboratory at the Tulane University Health Sciences Center in New Orleans.

Dr. Garcia's strong connection to Charcot-Marie-Tooth disorders and the Association began in 1987 when Dr. James Lupski from Baylor College of Medicine, contacted him regarding patients with CMT. They began a collaboration between Dr. Garcia's clinical work and Dr. Lupski's basic research, as well as beginning a long-term friendship. Dr. Garcia has followed approximately 280 CMT patients for over 20 years. The percentage of CMT patients in the MDA clinic at Lafayette is

exceptionally high—almost two thirds of the clinic patients have CMT. This high percentage of CMT patients has made the New Orleans area a prime source for families on which to do genetic studies.

The majority of the CMT patients in the New Orleans area represent the Cajun population, which has been stable for generations, making it easy to find the 4 or 5 generations of affected individuals necessary for involved genetic analysis. These French-Canadian descendants have been cooperative, friendly and gregarious. Dr. Garcia writes, "We had several field trips and cookouts at the homes of members of affected families. One person arranged to have his entire family—cousins, second cousins and all—come to the home during the day so we could do neurological exams, nerve conduction velocity studies, and blood draws for DNA analysis." Dr. Lupski was then able to take that information back to his lab for studies. "The collaboration and patience of these families was very important in helping us accomplish all the studies that we have done," stated Dr. Garcia.

In 1991, based on much of the information gathered in Louisiana, the duplication on chromosome 17 was discovered as the cause for CMT1A and the findings were published by Dr. Lupski, Dr. Garcia and others. In 1995, Dr. Garcia participated in the publication of *Charcot-Marie-Tooth Disorders: A Handbook for Primary Care Physicians* by writing the chapter entitled, "The Clinical Features of Charcot-Marie-Tooth Disorders." He has contributed articles to the newsletter and often serves as one of the doctors answering the "Dear Doctor" questions. He has served on the CMTA's Medical Advisory Board since 1990.



Over the past twenty years, Dr. Carlos Garcia has followed approximately 280 CMT patients.

Ask the Doctor

Dear Doctor,

I am a 46-year-old woman whose mother was diagnosed with CMT and had significant disabilities in her feet and legs early in life and later developed weaknesses in her hands and numbness and tingling in all extremities. She died about five years ago.

I was relatively asymptomatic until about seven years ago, when I started to develop tingling in my extremities, cold feet, and balance problems. However, for the past four years, I have also had significant swelling of the feet in the warmer months. I don't remember my mother having swelling of the feet until later in life, when she also developed some heart problems. Is swelling of the feet a typical side effect of CMT or should I investigate other causes for this problem?

A Member of the Medical Advisory Board (MAB) replies:

Peripheral neuropathy can cause swollen feet, but there are many, more common, causes. The lymphatics in the legs contain a series of one-way valves, and muscular contractions help to move the flow of lymph to the chest where it joins the venous blood. Thus, the lymphatic return is compromised by weak leg muscles.

I recommend that you see your personal physician to sort out why you have swollen feet.

Dear Doctor,

I changed my diet about seven months ago to eliminate meat and increase my carbohydrate consumption, while decreasing fat and protein. I currently get about 80% of my calories from carbohydrates and 10% from fat and protein. I started this diet to see if my energy levels would improve and so far the results have been very positive. I have more energy for exercising and am sleeping better. I lost about 10 pounds in the first few months and am now stable.

I have heard that people with CMT need meat protein and will lose muscle mass at a faster rate if they don't eat meat. My results would seem to contradict this, but I don't want to do damage over the long haul. Is it safe to eat a meatless diet?

A MAB doctor replies:

There is no literature that I am aware of that proves the need for "meat" protein rather than protein from other sources. As long as you have adequate protein intake and ensure that you are getting essential amino acids, you should be fine.

Keeping your weight under control and exercising adequately are two of the best things you can do for your CMT.

Dear Doctor,

Following several mentions of hyperbaric oxygen therapy (HBOT) on television shows, I began to wonder if it might be helpful for peripheral neuropathy, specifically CMT. I realize that CMT is genetic and that the actual neurological damage would not be reversed, but I wonder if the increased cellular respiration could possibly lead to a slowing of the progression of CMT or a decrease in some of the neuropathic pain symptoms I experience. Could HBOT, properly administered, cause an exacerbation of my CMT?

Several MAB doctors reply:

1) I am not aware of any cases of CMT which have been treated with hyperbaric oxygen. I know of no reason, other than cost, why such treatment would be harmful. However, I would emphasize that CMT disorders are chronic and it is hard to imagine how a brief period in a pressure chamber could provide chronic support. There is no evidence of which I am aware to suggest that there are reduced amounts of oxygen to either neurons or Schwann cells in inherited neuropathies.

2) Hyperbaric oxygen treatment (HBOT) is great for decompression sickness and to speed the healing of some infections. I do not know of any research done using HBOT in any type of CMT. I doubt that it would be helpful, although I do believe it would be safe.

3) I'm not aware of any treatments for CMT that included hyperbaric oxygen. I'm also not sure how such a treatment might help, especially in the case of the forms where we know the defect in a specific myelin gene causes the problem.

Dear Doctor,

I am a student of massage therapy and I have a friend who has CMT. She is 21 and all the women in her family have the disorder. Some of the symptoms I've read about are treatable with massage therapy. Are there any contraindications? Is there a type of massage therapy that should not be practiced on someone with CMT? Can massage therapy prevent the need for surgery?

A physical therapist replies:

Massage is often appropriate for people with neuromuscular disease. Because of frequent muscle strength imbalances, with some muscles becoming shortened and some becoming overstretched, massage can assist in maintaining muscle tissue mobility and thus aid in decreasing pain. I am not sure it could help prevent the need for surgery, unless only minor muscle restrictions are the problem.

As to contraindications, the most important is that people with CMT have sensory loss and abnormalities. A sensory exam should, therefore, precede any vigorous massage session or the use of heat/cold therapy.

**Dear Doctor,**

Does TES (threshold electrical stimulation) help with CMT disease at all? I have read anecdotal reports on two children in California mentioned on the home page who were able to give up their braces and had improved muscle mass on their lower calves. Do you have any evidence that this procedure does work for CMT patients?

A MAB doctor replies,

I am not aware of any medical evidence that TES is helpful in the treatment of CMT. Further, I am skeptical of stories about patients no longer needing AFOs or orthotic support due to TES. It is our feeling that good rehab medicine and an exercise program are likely to be more productive approaches to therapy.

PAIN

(Continued from page 7)

oxycodone, which lasts 3–4 hours). I typically ask patients to use a long-acting narcotic on a regular schedule (usually every 8 hours) to treat the constant pain, and a short-acting one for “break-through” pain (the episodes of pain that are not “covered” by the long-acting narcotic). When pain is worse during a particular time of the day (late afternoon to early evening), I recommend using oxycodone (for 2–3 hours of relief) or oxycontin/MS Contin (for 8 hours of relief), depending on the duration of that period. Like tricyclics, narcotics cause drowsiness, and have other side effects, too (constipation, for one). Tricyclics and narcotics often work well together for pain relief (tricyclics “potentiate” the pain relief of narcotics).

Tegretol (carbamazepine). This medication is not approved for the treatment of chronic pain but is probably more widely used for pain than for the treatment of its approved indication, epilepsy. Tegretol works by blocking voltage-gated sodium channels. In my experience, Tegretol works only sometimes. Tegretol comes in 100- and 200-mg tablets that are taken every 6 hours. A sustained released form (Tegretol XR) come in 100-, 200-, and 400-mg tablets; these are taken every 12 hours.

Anti-inflammatories. There are basically two kinds of anti-inflammatory medications—corticosteroids (these are different from the performance-enhancing “steroids” used by athletes) and nonsteroidal anti-inflammatory drugs (NSAIDs).

Prednisone, prednisolone, and decadron are examples of corticosteroids. These drugs are used for the long-term treatment of some chronic inflammatory conditions, but are more commonly used for short-term conditions. Corticosteroids should not be used to treat painful neuropathies, unless the underlying cause of the neuropathy is an inflammatory condition.

There are many NSAIDs, including:

- Aspirin, including Ecotrin. Aspirin or other salicylates are an active ingredient in many combination medications, including Excedrin and Disalcid.
- There are many newer NSAIDs—Anaprox/naproxen, Clinoril, Daypro, Feldene, Indocin/indomethacin, Lodine, Motrin/ibuprofen, Naprosyn, Orudis, Relafen, Tolectin, Toradol/ketoprofen, and Voltaren.
- The newest NSAIDs are the COX1 inhibitors—Vioxx and Celebrex.

Aspirin, and NSAIDs are not effective for the treatment of the pain of neuropathy, but they do work on radicular pain (caused by “pinched nerves”) as well as arthritis, tendonitis, and a host of other conditions.

CMTA Support Group News

■ California - Berkeley Area

The January meeting of the group featured a large turnout to hear Dr. Gail Widener, a physical therapist who stressed the need for an exercise program designed by a qualified therapist who knows Charcot-Marie-Tooth disorders.

The April meeting will be an opportunity to share those facets of people's lives that transcend the fact that they have CMT. Members are invited to come and introduce their hobbies, talents, crafts, or collections to others. Discussions about volunteer work, travel or family activities are also welcome. Those who wish to share their interests can, and those who prefer can look, listen and appreciate.

■ Kentucky/Southern Indiana/Southern Ohio

The March 24th program featured a psychiatrist, Dr. William Weitzel, who is in private practice in Lexington, Kentucky. He discussed the psychological aspects of living with an invisible, progressive debilitating disease. Patients were encouraged to forward questions to the doctor prior to the meeting so that the discussion could be relevant to the group.



CMTA President Ardit Fetterolf is co-organizer of the first meeting of the CMTA's support group leaders at the Frontenac Hilton in St. Louis, MO.

Support Group Leaders Meet in St. Louis For National Information Exchange

The first national meeting of the CMTA support group leaders will take place this April in St. Louis, Missouri. Approximately 18 of the group leaders, or their appointed representatives, will gather on April 27, 28, and 29 to discuss some of the strategies for making support groups better and keeping them functioning. Attendees will learn about what the CMTA is doing for its members and will offer suggestions about getting the information out at the local level about the CMTA and its projects. The new North American CMT Database will be discussed and the packets of questionnaires for the database survey will be available. Dr. Florian Thomas from St. Louis University will make a presentation on CMT and the research that is ongoing. A podiatrist will discuss the advantages of having shoes correctly fitted for a person's unique needs. Presentations will mix with sharing by the leaders and should result in each person taking back some new ideas and a new enthusiasm for the job of educating and serving persons and families dealing with CMT.

■ Missouri - St. Louis Area

The last meeting of the St. Louis area support group featured a presentation by member, Brenda Williams, on Paraquad, a community-based center for independent living. Both directors and staff members have disabilities, making insight into the ever-changing needs of individuals with disabilities their strength. They seek to combine direct services with advocacy initiatives that help shape public policy. Their goal is to remove not only physical barriers, but attitudinal barriers as well. Equal access to services, education, employment, transportation, housing and health care are on their agenda.

■ New York - Horseheads

The February meeting was the longest this group has ever had! It lasted almost 2½ hours. The speaker had to cancel because of an out-of-town emergency, but the members just discussed things of importance to those in attendance. One man drove three hours from Cobleskill, NY, just to be in the same room with people who understand his problems. He waited years for a correct diagnosis and talking with others who have CMT helped him with his frustration.

The group decided that they are helping each other with ideas, suggestions about medications, and the sharing of experiences—even bad ones.

CMTA Support Groups

Alabama/Greater Tennessee Valley

Place: ECM Hospital, Florence, AL
Meeting: Quarterly
Contact: William Porter, 205-767-4181

Arkansas—Northwest Area

Place: Jones Center for Families,
Rm. 206
Meeting: 3rd Saturday of each month
Contact: Libby Bond, 501-795-2240
E-mail: charnicoma57@yahoo.com

California—Berkeley Area

Place: Albany Library, Albany, CA
Meeting: Quarterly
Contact: Ruth Levitan, 510-524-3506
E-mail: rulev@pacbell.net

California—Los Angeles Area

Place: Various locations
Meeting: Quarterly
Contact: Serena Shaffer,
818-841-7763
E-mail: SerenaM71@aol.com

California—Northern Coast Counties (Marin, Mendocino, Solano, Sonoma)

Place: 300 Sovereign Lane,
Santa Rosa
Meeting: Quarterly, Saturday, 1 PM
Contact: Freda Brown, 707-573-0181
E-mail: pcmobley@home.com

Colorado—Denver Area

Place: Glory of God Lutheran Church
Wheat Ridge
Meeting: Quarterly
Contact: Marilyn Munn Strand,
303-403-8318
E-mail: mmstrand@aol.com

Florida—Boca Raton to Melbourne

Place: Upledger Institute,
Palm Beach Gardens
Meeting: Quarterly
Contact: Cynthia Gracey
561-243-0000

Florida—Miami/Ft. Lauderdale

Place: North Broward Medical
Center, Pompano Beach, FL
Contact: Al Kent,
954-742-5200 (daytime) or
954-472-3313 (evenings)
E-mail: marbearwld@aol.com

Kentucky/Southern Indiana/ Southern Ohio

Place: Lexington Public Library,
Northside Branch
Meeting: Quarterly
Contact: Robert Budde,
859-255-7471

Massachusetts—Boston Area

Place: Lahey-Hitchcock Clinic,
Burlington, MA
Meeting: Call for schedule
Contact: David Prince, 978-667-9008
E-mail: baseball@ma.ultranet.com

Michigan—Detroit Area

Place: Beaumont Hospital
Meeting: Three times each year
Contact: Suzanne Tarpinian,
313-883-1123

Michigan—Flint

Place: University of Michigan,
Health Services
Meeting: Quarterly
Contact: Debbie Newberger/
Brenda Kehoe,
810-762-3456

Minnesota—Benson

Place: St. Mark's Lutheran Church
Meeting: Quarterly
Contact: Rosemary Mills, 320-567-2156

Mississippi/Louisiana

Place: Clinton Library, Clinton, MS
Meeting: Quarterly
Contact: Flora Jones,
601-825-2258
E-mail: flojo4@aol.com

Missouri/Eastern Kansas

Place: Mid-America Rehab Hospital,
Overland Park, KS
Meeting: First Saturday bi-monthly
Contact: Lee Ann Borberg,
816-229-2614
E-mail: ardi5@aol.com

Missouri—St. Louis Area

Place: St. Louis University Medical
Health Center
Meeting: Quarterly
Contact: Carole Haislip,
314-644-1664
E-mail: c.haislip@att.net

New York—New York City

Place: NYU Medical Center/
Rusk Institute
Meeting: Monthly
Contact: Dr. David Younger,
212-535-4314,
Fax 212-535-6392

New York—Horseheads

Place: NYSEG Meeting Room, Rt. 17
Meeting: Quarterly
Contact: Angela Piersimoni,
607-562-8823

New York (Westchester County)/ Connecticut (Fairfield)

Place: Blythedale Hospital
Meeting: Monthly, Saturday
Contact: Kay Flynn, 914-793-4710
E-mail: alma622@worldnet.att.net

North Carolina—Archdale/Triad

Place: Archdale Public Library
Meeting: Quarterly
Contact: Ellen (Nora) Burrows,
336-434-2383

North Carolina—Triangle Area (Raleigh, Durham, Chapel Hill)

Place: Church of the Reconciliation,
Chapel Hill
Meeting: Quarterly
Contact: Susan Salzberg,
919-967-3118 (evenings)

Ohio—Greenville

Place: Church of the Brethren
Meeting: Fourth Thursday,
April–October
Contact: Dot Cain, 937-548-3963
E-mail: bobcain@wesnet.com

Oregon—Willamette Valley/ Pacific NW

Place: Alternates between Brooks
Assembly of God Church and
Legacy Good Samaritan
Hospital, Portland
Meeting: Third Saturday of the month
Contact: Jeanie Porter, 503-591-9412
Darlene Weston,
503-245-8444

Pennsylvania—Philadelphia Area

Place: University of PA, Founders
Building, Plaza Room A
Meeting: Bimonthly
Contact: Amanda Young,
215-222-6513
E-mail: stary1@bellatlantic.net

GIFTS WERE MADE TO THE CMTA...

IN MEMORY OF:

Mary Beeler

Bob and Barbara Allegro
McDonald's
Frank Rodegeb Family
Frank I. Rodegeb

Paul Budde

Bob and Phyllis Budde
Ellen Budde
Mrs. Josephine Budde
Neil Budde
Norman and M. Adine Budde
Harry and Dorothy Hinton
Naomi Paquinelli
Pittsburgh Presbyterian Church
Mr. and Mrs. Phillip Ritchey
Frank and Virginia Vonseggern
Joe and Marilyn Wunder

Muriel Fatigate

Kay Flynn

John Hill

Marilyn and Arthur Austin
Harold and Marilyn Averkamp
Roger and Jean Beckley
Mark and Ann Belsey
Roma Bolline
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William and Anne Conzemius
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Randy and Anna Saddison
Clayton and Susan Sager
Delbert and Nancy Schneeberger
William and Barbara Turner
Wolski's Tavern (Friends)

Andrew Hughes

Dr. and Mrs. Charles Bush

Carmine Pasquenza

Don and June Phillips

James Ryan

Jim and Kay Ryan

Brian Salley

TD Bank/Michael Bunga

IN HONOR OF:

Zohar Birin

Leon Gelman

Gail and Gaelynn Feeney Coyle

Mary Ellen Feeney

Jacqueline Donahue

Mr. and Mrs. John Reilly

Sheila Levine

Agents of Pacific Union

Eric and Michael Ponder

Margaret Drennan

Callie Marie Walden

Edna Casey

A. Hart Wurzburg's 80th Birthday

Mr. and Mrs. George Frank
Marjorie Grodin
Mary and Sam Lawton, Jr.
Mr. and Mrs. Joseph Lelewer
Jane and Robert Logan
Mr. and Mrs. Bud Minkin
Mr. and Mrs. Richard Simon

CMTA Remembrances

Your gift to the CMTA can honor a living person or the memory of a friend or loved one. Acknowledgment cards will be mailed by the CMTA on your behalf. Donations are listed in the newsletter and are a wonderful way to keep someone's memory alive or to commemorate happy occasions like birthdays and anniversaries. They also make thoughtful thank you gifts. You can participate in the memorial and honorary gift program of the CMTA by completing the form below and faxing it with your credit card number and signature or mailing it with your check to: CMTA, 2700 Chestnut Parkway, Chester, PA 19013.

Honorary Gift:

In honor of (person you wish to honor)

Send acknowledgment to:

Name: _____

Address: _____

Occasion (if desired):

- Birthday
- Holiday
- Wedding
- Thank You
- Anniversary
- Other

Memorial Gift:

In memory of (name of deceased)

Send acknowledgment to:

Name: _____

Address: _____

Amount Enclosed: _____

- Check Enclosed
- VISA
- MasterCard

Card # _____

Exp. Date _____

Signature _____

Gift Given By:

Name: _____

Address: _____



Letters to the Editor:

Dear CMTA,

I am a senior in high school who has CMT, and I am interested in studying how CMT affects decisions parents make with regard to having children and how they keep up with them if they do have some. I will face decisions about having my own children or adopting and I'm interested in how CMT patients make those decisions. Do you think children suffer if their parent has CMT?

—By e-mail

A member of the CMTA replied by e-mail:

I have a senior in high school myself and one who is a sophomore. I have CMT and was diagnosed when I was 16. I had high arches and always sprained my ankles, but I was busy climbing trees, riding bikes, etc. I had what I would consider a very normal childhood, other than the sprained ankles. I had surgery in my senior year in high school which slowed me down a bit, but once I recovered, I was back running, swimming, and playing softball well into my early thirties.

I married at age 32, had my first child at 34 and my second at 35. I ran after my kids after that. As an RN, I worked part time and full time plus being a wife and mother. I didn't know then what I know now about CMT. Would I make the same decisions? I don't know. I think I probably would. But, each person is different and CMT is different for each person. Some people run marathons and some are in motor scooters. It's a tough call. I think a genetics counselor could be helpful about the issue of having children or adopting.

Also, about keeping up with your children—it all depends on what you want to do with them and how your CMT affects you. There can be just as much closeness derived from sitting and reading together as in tossing a ball in the backyard. I'm in my early fifties now and I have noticed that fatigue is a problem, but I am still active. I don't run anymore but I do bike and walk. I can walk about a mile or two at a clip. I still work full time and I don't think my boys have suffered, overall, because of my CMT. We learn to adapt to the situation at hand. One can focus on the good things or the bad things. Most of the time, it's the good ones, but some days are better than others. That is true of everyone's life.

I hope this has helped and please feel free to ask any other questions.

By the way, my husband is supportive!

—K.H. by e-mail

Dear CMTA,

I thought I would pass along information on an item that has helped my father, who has CMT, and might help others who have muscle atrophy in the hands. My father can no longer turn on the switch for the floor lamp by his chair.

At the local hardware store, I found an item called a "Touch Dimmer" that plugs into your electrical outlet. You then plug the lamp into it, and by just touching the lamp, you can turn it on various light levels and then off. The lamp must be metal. This avoids having to buy a new "touch" type lamp and allows you to convert an existing lamp.

The manufacturer is Lamson Home Products, Cleveland, Ohio. Their home page is www.lamson-home.com.

—D.T., New Jersey

Dear CMTA,

My name is Nicole and right now I'm reading *Abby: Lost at Sea*. It is a great book! Abby has all of the same problems that I have and I can really relate to this book. I know that I have CMT, but I don't know a lot about it. My grandfather has it and I've had it for three years. I'm 13. I would love to talk to Pam Walls (the author) by e-mail.

(Editor's note: The request was forwarded to Pam, whose series of books with a heroine who has CMT was discussed in a previous issue of the newsletter.)

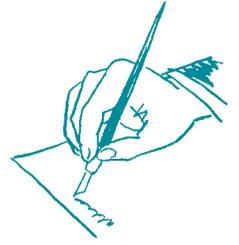
Dear CMTA,

Per chance, I stumbled on your article (Alternative therapies, a series of articles by Cynthia Gracey) on the CMTA website, while looking for more information. I am from Germany, but unfortunately, there is nothing like the CMTA over here.

The way your childhood was described is very similar to mine. I was born in 1968. CMT was diagnosed in 1980. In the next ten years, the disease progressed until by 1990, I was able to walk only for a few minutes at a time. I had foot surgeries in 1990 and 1991, and thankfully, I learned to walk again. Only a few weeks ago, I was told to stop my workouts because they could destroy what is left of my muscles and I was told the disease would get worse.

Anyway, I wanted to tell you that it's great that there's something like the CMTA out there.

—M.B., Germany



WRITE TO US!

Pat Dreibelbis, Editor

The CMTA Report

CMTA

2700 Chestnut Pkwy.

Chester, PA 19013

The CMTA reserves the right to edit letters for space.

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MEDICAL ALERT:

These drugs are toxic to the peripheral nervous system and can be harmful to the CMT patient.

Adriamycin
Alcohol
Amiodarone
Chloramphenicol
Cisplatin
Dapsone
Diphenylhydantoin (Dilantin)
Disulfiram (Antabuse)
Glutethimide (Doriden)
Gold
Hydralazine (Apresoline)
Isoniazid (INH)
Megadose of vitamin A*
Megadose of vitamin D*
Megadose of vitamin B6* (Pyridoxine)
Metronidazole (Flagyl)
Nitrofurantoin (Furadantin, Macrodantin)
Nitrous oxide (chronic repeated inhalation)
Penicillin (large IV doses only)
Perhexiline (Pexid)
Taxol
Vincristine
Lithium, Misomidazole, and Zoloft can be used with caution.

Before taking any medication, please discuss it fully with your doctor for possible side effects.

*A megadose is defined as ten or more times the recommended daily allowance.



What is CMT?

- ... is the most common inherited neuropathy, affecting approximately 150,000 Americans.
- ... may become worse if certain neurotoxic drugs are taken.
- ... can vary greatly in severity, even within the same family.
- ... can, in rare instances, cause severe disability.
- ... is also known as peroneal muscular atrophy and hereditary motor sensory neuropathy.
- ... is slowly progressive, causing deterioration of peripheral nerves that control sensory information and muscle function of the foot/lower leg and hand/forearm.
- ... causes degeneration of peroneal muscles (located on the front of the leg below the knee).
- ... causes foot-drop walking gait, foot bone abnormalities, high arches and hammer toes, problems with balance, problems with hand function, occasional lower leg and forearm muscle cramping, loss of some normal reflexes, and scoliosis (curvature of the spine).
- ... does not affect life expectancy.
- ... has no effective treatment, although physical therapy, occupational therapy and moderate physical activity are beneficial.
- ... is sometimes surgically treated.
- ... 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.
- ... Types 1A, 1B, 1X, HNPP and EGR-2 can now be diagnosed by a blood test.
- ... is the focus of significant genetic research, bringing us closer to answering the CMT enigma.

The CMTA Report

*Information on Charcot-Marie-Tooth Disorders
from the Charcot-Marie-Tooth Association*



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