

# THE CMTA REPORT

Providing information on Charcot-Marie-Tooth disease (a.k.a. Peroneal Muscular Atrophy or Hereditary Motor Sensory Neuropathy), the most common inherited neuropathy. Contents © 1994, CMTA. All rights reserved.

## Research Efforts Investigating Effects of Exercise in CMT

by Carol Oatis, PT, Ph.D.

The readers of this article know very well the common effects of Charcot-Marie-Tooth (CMT) Disease: muscle weakness and tightness, foot and hand deformities, difficulty in running or walking and, perhaps, difficulty with activities requiring hand dexterity or strength. These changes result from a progressive degeneration of the nerves that supply the muscles of the hands and feet. What is less well understood is the effect, both positive and negative, that exercise can have on these muscles and on the function of the person who has CMT.

Patients have been instructed frequently not to exercise and have been led to believe that exercise would stimulate the disease process. A variety of research studies published throughout this century do, indeed, describe a phenomenon known as overwork weakness. This weakness has been defined as a prolonged (but not necessarily permanent) reduction in strength in already weakened muscles following a period of work. Such weakness has been reported in people with polio and other peripheral neuropathies. However, most of these studies have been anecdotal in nature, lacking rigorous scientific controls. Similarly, animal studies have reported additional deterioration of the nerves in animals subjected to aggressive exercise following a nerve injury. These studies have made health care providers appropriately wary of exercise for people with CMT.

Yet, there also have been studies which have reported increases in strength and function in people with peripheral neuropathies following exercise programs. While few of these studies have actually looked at people with CMT, other disorders such as muscular dystrophy have been studied. Most of the studies which report positive effects of



Carol Oatis

exercise also report the need for caution in the application of exercise to avoid overwork.

Because of the controversy over the use and benefits of exercise, particularly in the person with CMT, we began a research program investigating the effects of exercise in people with CMT. The basic questions we are trying to answer are: 1) can exercise produce strengthening in already weakened muscles; 2) will exercise improve the function of someone with CMT; and 3) will exercise speed the degeneration of the nerves going to the exercised muscles?

Thus far, we have looked at the effects of exercise in two individuals with CMT. Each subject received a complete physical therapy evaluation. Then, we chose specific muscles to observe and specific tests to measure functional changes in the hands and feet. We also used simple electrical tests (not EMGs!) to observe changes in the nerves. We have been measuring these effects before, during and following an exercise regimen. Of

course, the results from only two subjects provide a very small piece of information needed to understand the role exercise may play in the treatment of CMT. These efforts, however, will be expanded to larger studies to enhance our understanding of exercise's effect on muscles weakened by CMT.

As we begin the third year of this study, our results have not been published in any professional journal although we will be submitting one article in the next two months. Publication is essential in order to allow health care providers and researchers to scrutinize and challenge our findings. Only after such scrutiny is it reasonable to present them to the general public. Consequently, it would be

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## Summary

### *CMTA Medical Advisory Board Meeting in Washington, DC*



*Michael Bennett discussed connexin and its role in CMT X.*

In May, at the meetings of the American Academy of Neurology in Washington, DC, members of the CMTA Medical Advisory Board, chaired by Dr. Robert Lovelace of Columbia P&S, were brought up to date on recent and exciting research which has found CMT gene locations. This mapping is the necessary first step toward identifying the gene product. Only after the gene product is determined can the possibility of replacing altered protein be addressed and real treatments for CMT begun. A number of laboratories and inspired workers in CMT research have led to this progress.

Dr. Thomas Bird of Seattle, who in 1980 was the first to link CMT I with chromosome 1, chaired the meeting. He outlined the latest classification of CMT genes. The most common form, CMT IA (dominant inheritance, slow nerve conduction) gene is found on chromosome 17. Alterations in the gene include gene duplication, deletion, and trisomy. This locus is associated with point mutations of the pmp22 protein, a protein of peripheral nerve myelin. Strangely, a similar but different condition, Hereditary Neuropathy with Liability to Pressure Palsies, described by Drs. Philip Chance of Philadelphia and Tom Bird, is also located on chromosome 17. However, in HNPP, pmp22 deletions occur. Gene abnormalities on chromosome 17 can now

be identified by a blood test developed by Genica Pharmaceuticals of Worcester, MA.

The CMT Ib gene, a less common CMT form, but one still having a dominant inheritance and slow nerve conduction, is found on chromosome 1 (Dr. Roger Lebo, U of CA). Here the abnormal gene product appears to result from point mutations in the PO myelin protein. No test is available for this gene. In addition, researchers believe that there are further undiscovered CMT genes as there are dominant CMT families who do not have abnormalities at the known chromosome 17 and 1 locations.

Other CMT genes were discussed, including the gene on the X chromosome, which is clearly shown in many families to have at least 2 gene loci. Dr. Victor Ionasescu of Iowa and others have mapped CMT genes at Xq11-21 and Xq26.

The CMT IIA gene, is thought to be on chromosome 1. This less common form of CMT has been more difficult to study. Dr. Jeffrey Vance described CMT IV A, a CMT form present in Tunisia. He located this gene on chromosome 8, but has not yet learned the protein gene product.

The final speaker was Dr. Michael Bennett, Albert Einstein College of Medi-

cine. Dr. Bennett's lecture gave a fascinating new insight into nerve structure and function. One CMT X chromosome deletion appears to interfere with the function of a protein called connexin 32 (C32). This protein is an important part of gap junctions of Schwann cells, the cells which make peripheral nerve myelin. Gap junctions are small complicated water-filled channels that connect the insides of neighboring cells, permitting the passage of chemicals and electrical currents between them. Dr. Bennett suggested that when the gap junction channels were not functioning properly, the most distant Schwann cells would not receive proper nutrition and/or stimulation. They might be unable to make myelin and eventually might die. This would result in demyelination or myelin loss, the common defect in CMT peripheral nerves. There are still many questions, such as the presence of C32 in many organs that do not seem to be affected by CMT, and the multiple forms of C32 that have just been discovered. However, the connexin link to X-linked CMT is an exciting clue in understanding myelin loss.

The researchers present praised the CMT families who have participated in all of these and other studies. While they agreed that much work still needs to be done, the CMTA Medical Advisory Board wants the CMT patient to know that many medical centers are involved in active CMT research. Together, they hope to reach the common goals of understanding and treating CMT.

*Lowell L. Williams, M.D.*

*Editor's note: Dr. Otis' research results will be published in the CMTA Report as soon as they are available.*

#### **Exercise & CMT- cont'd from p.1**

irresponsible of us to present even preliminary findings in this newsletter. However, we can tell you that our research has generated considerable interest among physical therapists and physical therapy students. This interest alone will benefit those people with CMT. As our findings are reviewed by other investigators, we will continue to make those findings available to you. §

# Items of Interest

**Item 1:** A book called Colleges that Enable by Jason and Prudence Tweed investigates more than 2,000 college campuses and offers the following nine colleges as having the best services for students with mobility impairments:

- Edinboro University, PA
- Louisiana Tech University Ruston, LA
- St. Andrew's Presbyterian College Laurinburg, NC
- University of California Berkeley, CA
- University of Houston, TX
- University of Illinois at Urbana Champaign, IL
- University of Kentucky Lexington, KY
- University of South Carolina Columbia, SC
- Wright State University Dayton, OH

**Item 2:** A patient recommends the following book: Living with Chronic Illness: Days of Patience and Passion by Cheri Register. The book is published by Bantam, NY, 1987. The ISBN number for ordering from a bookstore is 0-553-34764-0. The patient calls this book "a support group in a book."

**Item 3:** There is a new quarterly magazine called "Common Journeys." It is a participatory journal for and by readers living with chronic pain or illness, as well as for those in a caregiving or family role. "Common Journeys" can be reached at P.O. Box 17003, Minneapolis, MN 55417, (612) 729-7552.

**Item 4:** In the last issue of the *CMTA Report*, we announced a new program enabling CMT patients to donate tissue, before or after death, for research. Those people who have contacted the CMTA have been sent the information. In the next issue of the *CMTA Report* we will publish a full account of the program. If you are interested in further information about this, write:

The CMTA Gift of Hope  
601 Upland Ave.  
Upland, PA 19015

## Prenatal Testing for CMT

*A.E. Harding, MD, FRCP,  
Institute of Neurology, London, England*

The rapid recent progress in research has led to the identification of several genetic abnormalities underlying different types of CMT. This means that it is now theoretically possible to test people from CMT families, including prenatally, to see whether or not they have the genetic abnormality which causes CMT in their family. At the moment, this largely applies to families with the duplication of chromosome 17 seen in most families with CMT 1 (CMT 1A). I will focus in this article on prenatal testing in relation to CMT 1A.

Anyone with CMT 1A has a 50:50 chance of passing it on to each of their children. The condition does vary in severity, ranging from causing no problems at all to causing major difficulties in walking in early adult life; the latter is unusual. Before even considering prenatal testing for the duplication, it is essential to confirm, in advance of the pregnancy, that the prospective parent has the duplication. There is no point at all in testing for the duplication in families who have other sorts of CMT.

Personal experience with many CMT 1A families suggests that requests for prenatal testing are infrequent; we have only been asked to do this twice in London and we provide a laboratory service for most of the UK. Couples' views on this

issue will inevitably be coloured by their experience with CMT, either in themselves or their relatives. Many feel that termination of an affected fetus is not justified by the (usually) relatively mild disability of CMT 1A. It is impossible to predict from the test whether the individual will be severely or mildly affected. However, some couples cannot countenance having an affected child, with about a 20 percent probability of being significantly disabled in adult life, and will opt for prenatal diagnosis with termination of an affected fetus. The prenatal test is done on a chorionic villus (afterbirth) sample, taken with a fine cannula through the neck of the womb at about 10 weeks after conception. The test needs to be done by an experienced laboratory using a reliable and validated technique. It can be done later in pregnancy, but this means that if the pregnancy is terminated, it is more unpleasant for the mother as she has to go through a mini-labour. Testing should not be done unless the couple concerned is as sure as possible beforehand that they will terminate an affected pregnancy. There is nothing to be gained in knowing whether or not the fetus has the duplication for any other reason, and potentially much to be lost. Chorionic villus sampling has about a 1 in 50 risk of causing miscarriage. §



### CMTA Chapter Development

-By Diana Eline

Do you have talents in Leadership, Publicity, or Fundraising or would you simply like to help? Would you like to organize a local Chapter of the CMTA or help in getting one off the ground?

The CMTA is ready to begin Chapter Development, which would lead to a stronger presence of the CMTA in local areas, and new opportunities for public awareness, fundraising, and getting to know others with CMT.

If you have an interest, please call/write the CMTA, 601 Upland Ave, Upland, PA 19015, 610-499-7486.

### Call for Articles

*The CMTA Report* welcomes your ideas and article suggestions. For example, you may submit a human interest story telling of your experience of living with CMT. Also, medical professionals can forward articles of a clinical or medical nature that would be of general interest to our readership.

### Just the fax.

Is your office upgrading and replacing your current fax machine? We have a need for an auxiliary fax machine to be used off site. Of course, the donation would be a charitable contribution.

# A Family Reunion with a Serious Twist

## *The Mattingly Reunion*

by Andrew Mattingly Jackson

Like many family reunions, the Mattingly family get together in Calvert County, Maryland, allowed people who rarely see each other an opportunity to share stories, food, and fellowship. What was unique, however, was the underlying purpose for the gathering.

Masterminded by Andrew Mattingly Jackson, the reunion brought together 150 members of the Mattingly family along with medical researchers from Johns Hopkins and the Children's Hospital of Philadelphia (CHOP). The purpose of the gathering was to allow the scientists to obtain blood for DNA testing and medical histories from a large number of Mattingly descendants, including the healthy ones. By comparing DNA from afflicted and non-afflicted family members, the researchers will try to isolate the defective gene to determine what causes the breakdown of information from brain to muscle, resulting in muscle atrophy.

Researchers at the National Institutes of Health (NIH) first discovered the Mattingly family problem in the 1960's and diagnosed the ailment as Charcot-Marie-Tooth. Now, however, the doctors believe that the Mattinglys suffer from a variant of CMT. It currently has no name and no cure. Unlike CMT patients, the Mattinglys retain feeling in their fingers and toes. Moreover, the family disorder seems to be more severe than CMT leaving several family members, including Andy Jackson, dependent on wheelchairs for mobility. Andy has also experienced such severe muscle wasting in his hands and arms that he is unable to feed himself.

However, none of Andy's physical problems kept him from planning and executing an amazing gathering that excited the medical research community and provided an opportunity for parents and potential parents to ask questions about the family malady.

The CMTA salutes Andy and stands in awe of this remarkable gentleman. The following is Andy's account of the reunion. §



*The organizer, Andrew Jackson Mattingly, with the youngest attendee, his grandson Andrew Jackson Mattingly IV*

In the hopes of keeping the disease, that affects the Mattinglys, from crippling future generations we held a reunion of all persons who trace their roots to the Mattinglys, who came to St. Mary's county (MD) from Mattingly England, during the 1600's. The dates were June 4th and 5th, 1994, and the location was Solomons, in historic Southern Maryland.

We had this gathering for both social and scientific reasons. From the social aspect such a gathering gave many of us the opportunity to meet relatives we have not meet before. From the scientific point we hope to find the genetic cause of the motor neuropathy, which prevails in the Mattinglys and their descendants. The information gathered at our reunion will be shared with other scientists who are studying neuromuscular diseases.

In attendance were descendants of Thomas Mattingly, who died in 1664; and descendants of George Washington Mattingly, who was born in 1821 and died in 1871. Also present were Drs. David Cornblath, John Griffin, and Bruce Rabin from Johns Hopkins University School of Medicine, and Drs.

Philip Chance and Mena Scavina from University of Pennsylvania School of Medicine.

Researchers at NIH examined a number of Mattinglys, including myself, in 1963. The disease was diagnosed, at that time, as Charcot-Marie-Tooth. While the disorder in the Mattingly family has been called CMT for generations, re-examination of that concept has suggested that indeed it may be a unique disorder in our family and not CMT. We have sensory feeling in our extremities, and nerve conduction velocity in some of us who have been tested is somewhat normal. Also the disabling affect in many Mattinglys is more severe than most people with CMT. It is the opinion of the neurologists who were there that this Mattingly degenerative neuro-muscular disease is a form of amyotrophic lateral sclerosis, but not the type that affects the muscles in the internal organs. Also, life expectancy is not shortened by this disease.

In order to find the mutated gene responsible for the malady and to help find the cause and the cure of disease, it was first necessary to develop a family tree. A

twelve foot long family tree was developed and posted on the wall of a ballroom at the reunion. This showed those persons with the disease, starting with George W. Mattingly, who died in 1871. In addition an 80 page "Mattingly Family Genealogy" chart was prepared. This started with Thomas Mattingly in 1664. Every family that attended received a copy of the chart.

Family members were examined by the team of neurologists who were at the reunion. Those who fit the following criteria had their blood drawn:

- Affected individuals their spouses, children, and parents.
- Unaffected, at-risk individuals, as defined by the disease being present in either a parent or sibling.
- Unaffected obligate carriers.

Blood was not taken from the spouse or children of clearly unaffected individuals.

The blood samples were taken to CHOP by Dr. Chance, where they will undergo specialized genetic testing; to determine which chromosome carries the degenerative neuromuscular disease trait; to find out what exactly the disease trait is; to determine how it causes the disorder; and to determine how it can be treated. Both the examination and drawing of blood were done by appointment. This gave the participants the opportunity to sightsee and enjoy the amenities at the resort.



Dr. Philip Chance addressing the Mattingly family

For those who live out of town or could not attend, special arrangements will be made to have them go to a laboratory near them to give blood, and to ship to Dr. Chance for testing.

The reunion was a huge success from both a social and scientific standpoint. From the social aspect, many of us had the opportunity to meet relatives we had never seen before. Also there was a certain amount of comfort being with peo-

ple, other than those in your immediate family, who are in the same situation as you. In addition, the fellowship and conversations during the cocktail party, the dinner, and the after dinner comments by Dr. Chance were so pleasing that no one wanted the day to end.

Approximately 120 people were examined, and 80 blood samples were taken. These samples, along with 8 other samples previously taken, are at CHOP. About 25 more blood samples will be given by people who were unable to attend the reunion.

Because of the efforts on the part of the CMTA, the reunion received newspaper coverage by The Washington Post, The Baltimore Sun and the Associated Press. The AP put a story over their wire service that was picked up by other newspapers who published the story.

*Editor's note: Andy is interested in hearing from other people who have similar symptoms. He may be contacted at:*

400 Houcksville Rd.  
Hampstead, MD 21074  
or by calling 410-239-8899.

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## ¿ Habla usted Español?

The CMTA announces the publication of a Spanish edition of our CMT Informational brochure (gray brochure). For a copy check the block on the order form on page 15. For multiple copies contact the CMTA office.

Do you know of anyone famous who is a CMT patient? Do you know anyone famous who would advocate for CMT? The CMTA is looking for a well-known person to be a spokesperson for CMT. If you know of such a person, contact the CMTA; we will do the rest.



Socializing at the Mattingly family gathering in Maryland



## Clinical CMT Research Update

There have been two recent CMT articles in scientific journals that are of great interest to the CMT community and the medical community. We learned about the first article from Dr. Thomas Bird, the principal author. It appeared in *European Neurology*, 1994;43:155-57 and is entitled "Impotence Associated with the Charcot-Marie-Tooth Syndrome". In the article's introduction the authors state, "Impotence is a fairly common complication of numerous polyneuropathies. The best example is diabetes mellitus. Surprisingly, there has not been a formal report of impotence associated with the Charcot-Marie-Tooth syndrome (CMT; hereditary motor sensory neuropathy, HMSN), even in extensive reviews of the disorder. We report here 7 instances of this association and suggest that it is more common than previously recognized." For a copy of this article write to Dr. Thomas Bird, Neurology Service, VA Medical Center, 1660 South Columbian Way, Seattle, WA 98108.

The second article appeared in *Foot & Ankle*, Vol. 14, No. 8/Oct. 1993 and is entitled "Foot and Ankle Manifestations of Charcot-Marie-Tooth Disease", by James R. Holmes, MD and Sigvard T. Hansen Jr., MD. This is a very comprehensive discussion of the CMT foot and we feel that it has great value for the patient and physician. Therefore, we are printing the authors' conclusions as written. (*Editor's note: The following information is written for physicians and contains technical words and phrases. For clarification of anything you do not understand, take the article to your physician for his/her explanation.*)

**Conclusions** "Orthopedic management of the foot and ankle problems associated with Charcot-Marie-Tooth disease continues to evolve. While disagreement persists as to preferred treatment options, we believe the following statements are consistent with current understanding of this complex problem:

1. "CMT disease" should be considered a spectrum of neurological dysfunction with variable inheritance, clinical course, and severity of involvement.
2. Over one half of the patients with CMT have foot and ankle difficulties that may include weakness and/or paresthesia, pain, deformity, or an unsteady gait.

3. The typical foot posture of CMT is that of cavovarus deformity with predominantly anterior cavus. While some controversy exists as to the pathomechanics, a relative imbalance between (1) peroneus longus and tibialis anterior and (2) tibialis posterior and peroneus brevis has been observed and is likely a chief contributor to the pathologic foot posture.

4. The foot in CMT must carefully be examined to determine the identity and magnitude of the deforming forces, joint instability, and the flexibility of the deformity. Treatment must be individualized based on these findings.

5. Nonoperative treatment, including night splints, ankle-foot orthoses, shoe modifications, and other orthotic devices, has been relatively unsuccessful in treating significant and/or progressive foot deformity.

6. A variety of soft tissue procedures have been advocated and are most useful for correction of flexible deformity, especially in younger patients.

7. Many midfoot osteotomies have been described in the treatment of pes cavovarus deformity secondary to Charcot-Marie-Tooth disease. However, if a rigid, severe deformity is present, triple arthrodesis seems to have emerged as a more popular bony procedure.

8. Results of triple arthrodesis for CMT deteriorate with time and are considered by many authors to be a salvage procedure in this disorder.

9. Results of treatment in CMT foot problems are difficult to interpret because of the wide spectrum of neurological dysfunction, deforming force characteristics, degree of stiffness, and surgical procedures described. However, it appears that relatively early intervention (in the flexible foot) consisting of soft tissue releases, tendon transfers, and, perhaps, nonarticular osteotomies not only restore more normal foot biomechanics and posture, but may help prevent or delay the need for more extensive bony procedures."

There is a 52 item bibliography with this article. For a reprint write to: Dr. James R. Holmes, Orthopedic Surgery Associates, 5333 McAuley Dr. #R2009, Ypsilanti, MI 48197. §

## Gene for Neurological Disorder Identified by MSK Researchers

(This article appeared in the *Center News*, a publication of the Memorial Sloan-Kettering Cancer Center and is reprinted here with permission. The article is copyrighted.)

Dr. Jeffrey V. Ravetch and colleagues in MSK's Molecular Biology Program have identified a gene that, in its altered form, causes Charcot-Marie-Tooth (CMT) disease, a severe neurological disorder that afflicts more than 100,000 Americans. "This discovery could lead to prenatal diagnosis of this disease as well as the development of drug treatments," said Dr. Ravetch. The research grew out of his long-standing investigation of genetic control of the immune system.

CMT disease was named for the three French and British physicians who first described it in the 19th century. People with this disorder typically experience problems with movement and sensation in the lower legs, feet, forearms, and hands. The muscles in these limbs atrophy, leading to severe weakness. It can also affect the muscles used for breathing. The disease arises from defects either in nerve cells or in myelin, the fatty sheath that surrounds nerve cells and plays a significant role in the conduction of nerve signals.

The form of CMT disease being studied by Dr. Ravetch, type 1B, had been known for several years to be caused by a defect

(continued on next page)

### Call for CMT-X Participants

If you are a CMT-X patient or a member of a CMT-X family, Dr. Michael Bennett, chairman of the Neuroscience Department at Albert Einstein College of Medicine, needs you! Dr. Bennett is conducting research on connexin 32, the malfunctioning chemical compound in CMT-X patients. If you have the diagnosis of CMT-X in your family, contact Dr. Bennett at the Dept. of Neuroscience, Albert Einstein College of Medicine,

# On College and Being Disabled

by Jean Meyers Ryan

Last fall, after 25 years in the "real world," I started graduate school. I have CMT and vision problems. Included with the application form was another paper mentioning the disabled-student-services office. Although I didn't know what they offered, I thought I could do fine without them. After all, I thought, there were probably people in wheelchairs or who were blind, who really needed those services. Was I ever mistaken!

As the weather got colder, I discovered that I couldn't carry a winter coat and my briefcase in one arm while I held my cane with the other as I walked to classes. Once, while trying to get out my student ID for entrance into the library, I accidentally dropped everything at the guard's entrance to the library. I was occasionally "wiped out" with pain and fatigue. At the final exams, my concentration was adversely affected by blurry vision from the bright fluorescent lights. I signed up right after the test!

Before the second term began, I was invited to a disabled students' orientation. The counselors addressed the issues of notifying professors at the beginning of the term that we are disabled. Will it help or hurt? Why are we entitled to double time on tests, notetakers, readers, extended deadlines, or lockers for our coats and books? Here is their rationale, which makes sense to me:

There may be unforeseen health problems which develop as the term or year goes along. Already having a working relationship with your professor and disabled student services is like having insurance. You may not need it, but if you do, it is available immediately. Imagine the scenario in which you have not identified yourself as disabled and you experience pain and fatigue the weekend before your 25 page research paper is due. How will the professor take your request for extra time when you ask for it on the due date?

Sometimes you can anticipate difficulties and plan around them in advance. For example, I will be student teaching in a year-and-a-half. I know I will not be able to go to a school every day and travel 2 hours each way. I can no longer do that. I already notified my program chairman that I will have to student teach over two terms and go in 3 days a week. I also need a school which is accessible and has ele-



Jean Meyers Ryan

vators. (Most New York City schools are 4 or 5 stories and only have steps.) My chairman first said, "No one has ever done that before." I smiled and said, "I'm sure it can be worked out," all the while thinking that I'm certainly not the only disabled person in New York City to have to split my student teaching over two terms, but I'm probably the first in his department. I also went to see a dean who is assigned to helping disabled students get accommodations. Each term, I will check on the progress of the plan. I will have a better chance of doing the student teaching this way than if I tried it and got too tired after a week.

It is no fun to have the professor know right away that you are disabled and stand out from other classmates. We all want to be known mainly for our qualities. However, the professor is legally not allowed to use your disability against you or refuse to make reasonable accommodations for you. Most professors want to help, but they don't know how. Usually the uncooperative professor can be educated by the student and student services about the need to cooperate and, if it comes to that, his/her legal responsibilities. If you encounter a problem with a professor, you can get help resolving it from student services, the heads of that department, or a dean. This works in any college or university.

If you are really fatigued and in pain, or if you have blurry vision or trouble writing fast or at all, you might not be able to do well on a test in the normal amount of time. Then the test would be testing your disability and not your knowledge. If you have extra time for a test, you will not do well if you do not know the material. The same argument goes for some deadlines you may not be able to meet for written assignments.

It is up to us to take advantage of services which can actually help us to be more productive and effective. "Toughing it out" alone is not necessarily the solution. It may not be possible. Sometimes "toughing it out" is doing the work, learning the material, finishing the term, and passing the course. Accommodations are not cheating; they are not letting a student get away with less work. They are helping a disabled student to do as well as other students by not letting the disability get in the way.

*Editor's note: Jean Myers Ryan's philosophy is certainly uncommon good sense.*

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## Gene for... cont'd from p. 6

in some unidentified gene on chromosome 1. Dr. Ravetch's team, which includes David G. Brooks, an M.D./Ph.D. student working in his laboratory, has now reported that type 1B is caused by a defect in the gene responsible for triggering the production of a protein that is a major constituent of the myelin sheath. When the gene is altered, cells produce a defective form of the protein that reduces the myelin's ability to function. As a result, nerves cannot carry messages from the brain to the muscles, and the muscles atrophy.

Dr. Ravetch's main line of research concerns immune-system receptors that are involved in the recognition of antigens and antibodies. The genes for these receptors are also located on chromosome 1. When Dr. Ravetch learned that the gene for type 1B CMT disease was believed to be in the same region, he launched a search for the gene. In collaboration with Dr. Roger V. Lebo of the University of California at San Francisco and Dr. James A. Trofatter at Massachusetts General Hospital, genetic probes - short stretches of DNA that attach to genes associated with a disease - were developed by Dr. Ravetch and were used to locate the gene.

Dr. Ravetch is now developing a laboratory model that will have the genetic defect and thus can be used to screen potentially helpful drugs.

His work was funded by the National Institutes of Health.

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# CMT and Me

by Mary Elizabeth York



Mary Elizabeth York

The little girls sat against the side of the room, hands folded primly in their laps, eyes downcast, each wondering which boy would ask her to dance. Then the boys came; each bowed to the girl of his choice; and two by two they went on to the dance floor until there was but one boy and one girl left. The boy looked around, not wanting to ask this girl sitting hopefully waiting, but there was no other choice. Selfconsciously, she arose to dance. As she danced, she stumbled frequently, blushed, apologized and tried again. Somehow, she couldn't seem to get the steps as fast as the others; her brain understood them, but her feet just wouldn't follow, and a wrong turn would throw her off balance.

"Why," she wondered, "Why?" "Am I going to be lame like Mother?" Her mind went back to the time her mother, who always walked with a cane, fell in the middle of a busy street. How panic stricken she had been! She thought, too, of the many times she'd waited to be chosen to play baseball, and had always been left until last because she couldn't run fast enough. And of the times the scout counselors had waited for her on hikes and helped her over the rough ground. Certainly, she wasn't a pleasant dancing partner when she was so miserably self-conscious. The boy was quicker the next time; he didn't get stuck with her again.

As the girl grew up, she learned to compensate for this disability. Physical education was discarded in favor of the glee club. As the years went by, she grew out of her self-consciousness... Her philosophy of life was directed toward putting her talents to active use and learning to live with her disability.

Under the title, "There is No Cure," this is what I wrote about myself in 1953 when I was 32 and taking a class in composition at Arizona State University. Only a year or so before, a neurologist

had attached the name, "Charcot-Marie-Tooth" (CMT) to my disability. Not much was known about CMT then. Even today, 40 years later, not much more is known. There is still no cure although genetic research shows some promise.

That my disability was inherited was clear. I remember walking to the trolley stop to meet my grandfather coming home from his office. He walked with a cane throwing his foot forward with a slap. In a family history, Grandfather wrote. "My father, James Nellis had married Mary Magdalene Wert in Johnstown, January 25, 1842. They were both singers and had met in singing school. She was the daughter of Daniel Wert and Hannah Coughnet who were full cousins... Both were lame in their last years. Their families before them had twice intermarried and future generations should take warning." Elsewhere in family history, it mentions that Mary Magdalene was lame also. A family legend tells of a coat of arms showing a hand with the thumb extended at right angles to the palm and a crooked little finger. I have never seen it and so do not know whether or not it exists.

I do not remember my mother ever walking without a cane. With the years, she became progressively worse and her feet became very deformed so that the toes curved inward and she walked on the outside of her feet. By the time I wrote the above composition, she was 71 and had used a wheelchair for several years.

As for myself, I did not walk until I was three. Mother said I would hitch myself around in a little rocking chair. When I did walk, tiny steel braces were made for me to hold my feet straight, and I wore high-topped shoes... much to my disgust... until I was ten. As children do, I bragged about being able to bend my thumb back against my wrist... double jointed we called it... not realizing it was part of the weakness. I was never able to ice skate or wear high heels; my ankles were much too weak. It was, however, a minimal and essentially invisible handicap with attendant pluses and minuses. The handicap didn't keep me from living a normal active life, marrying and having children and then attending college and becoming a teacher. On the other hand, a supervisor wrote an evaluation of my teaching and criticized me for not wearing heels... a criticism she wouldn't dare have made after the civil rights movement.

My daughters had no sign of CMT, but my son did. We went to an orthopedic surgeon who put us both in braces to keep the heel tendon stretched. As my son kept breaking his in his active play, which included climbing trees and jumping out of them, we discontinued their use. Nor did I think mine were helpful.

By my early forties, my toes had curled under so that I walked on the tips, a condition so painful I could walk scarcely a block unless I was heavily dosed with aspirin. In 1964, I had foot surgery in which the heel tendons were lengthened and the toe joints fused. This was so successful that I walked relatively easily for about 22 years. Gradually, however, I fell more frequently, tripping over a toe which, instead of being lifted normally as I stepped, hung limply downward. I was referred to an orthotist by a physiatrist and, in 1986, a brace was made to keep the front part of the foot at right angles to my ankle.

In 1993, I learned of the Charcot-Marie-Tooth Association and immediately joined. I am grateful to know that I am not alone and that there are health professionals who know what CMT is. Too often, I encounter doctors who know little or nothing about CMT, or if they do, simply say that there is nothing to be done about it. They seem unaware that surgery can help or that exercise is important. I have found both helpful. In addition to the foot surgery, I had hand surgery in 1989 bringing my left thumb into line through a tendon transplant. Now I can use that hand to button buttons, something I had lost the ability to do. I believe that my swimming regularly and my son's bike riding, as well as exercises prescribed by physical therapists, have helped us to avoid the decline that my mother experienced.

Each case is different even within our family. My feet are relatively straight while I have difficulty with my toes; Robert's toes are straight while his feet tend to twist inward as Mother's did. My brother also had CMT, but didn't realize it until late in life after he had acute rheumatoid arthritis.

I allowed my name to be used in the CMTA newsletter as a contact person for a support group in Oregon, and have received two phone calls. Our hope is that others will join us and we can indeed form a support group and learn more about CMT. §

## Patient Profile

# Paul "Go-Go" Gomez

by Diana Eline

Paul Gomez is someone we can all learn from. He deals with his CMT with a certain acceptance and a strong sense of humor. At 11, he was chosen by the Muscular Dystrophy Association first as the local goodwill ambassador (formerly referred to as Poster Child) and later for the entire state of New York. His spirit of cooperation and his ability to relate to others have both helped to earn him his title.

CMT affected Paul at a young age. He attends 5th grade with all the other kids at Brewerton Elementary School in Brewerton, New York. However, due to severe hand problems he has an aide provided by the school who helps him every day with writing and other tasks that he is unable to perform. Paul also has to get up earlier than the other kids his age. Before the regular school day begins, he participates in physical and occupational therapy also provided by the local school system. He takes a special bus to school as he requires help getting onto the bus. He has worn Anterior Foot Orthoses (AFO's) since the age of 4 to aid him in walking. Presently, for long distances he relies on a wheelchair. If Paul falls down, he lacks the muscles to get himself back up again. His arms are also affected. He has difficulty in raising his arms above his head and is unable to open large doors. Again, because of his hand problems he needs help in dressing and grooming.

Whereas Paul is unable to excel in physical activities, he does very well in academics. He said that he would like to be either a teacher, a lawyer, or a comedian when he grows up and added that he wouldn't mind going to Yale. Besides, he adds, he already owns a Yale sweatshirt.

When asked what he is the most proud of, he said his ability to ride roller coasters without any fear. He has mastered 15 different roller coaster rides at his favorite amusement park, Kings Island, in Cincinnati, where he goes to visit relatives. He also enjoys hobbies such as rock collecting and fishing.

Despite his obvious disabilities, Paul always manages to make friends. He says that in school, in order to avoid instances of kids poking fun at him, he often makes friends with the biggest and strongest boys in his class who, in turn, won't let



Paul Gomez

anyone bother him. He also has several good friends in his neighborhood. They have even modified the game of football so that Paul can play. When he walks with the football, penalty points are given if anyone knocks him over.

Paul's dad Ken, and his sister, Erica (15) also have CMT, but are significantly less affected than Paul. Along with his mom, Sandra, they help Paul when he needs help and encourage his abilities.

As the MDA ambassador, Paul will spend the next two years attending MDA fund-raisers and events and will be involved in the telethon in September. It takes special people like Paul and his family to make such a strong commitment.

I had a wonderful experience in meeting Paul. I have also learned a great deal from him. Despite his difficulties, his charm and wit dominate. His effort to be as involved and active as possible is remarkable. It's no wonder his friends call him "Go-Go" Gomez. §

**CMTA  
Conferences  
coming soon to  
Philadelphia and  
New Orleans...**

**It's a great opportunity-  
Don't miss it!**



## Ask the Doctor

Dear Doctor,

What proportion of CMT people have muscular weakness in the lower back, which affects stair climbing? What can be done about it, if anything?

What proportion experience muscular weakness in the shoulders? I find my swimming is being restricted by that.

What proportion have paresthesia in the shoulders? I have experienced some in the left shoulder and back, but it seems to be easing now. About ten years ago, before my CMT was diagnosed, a chiropractor said he could not figure out why he could not finish straightening out my back. (Now I think it is the CMT.) In other words, doesn't CMT affect all the muscles of the body, to varying degrees, but especially the extremities of the hands and feet?

I would appreciate whatever you can tell me about this, which will let me know what to expect in the future.

Brooklyn, NY

**The Doctor replies:**

*The weakness that occurs in CMT is usually distally in the legs, and this affects walking and climbing in many patients. Low back pain, weakness, and fatigue may be secondary to the lower extremity weakness, but may reflect musculoskeletal problems of the spinal column. This can be evaluated by neurologic examinations, spine xrays and imaging studies, and electromyography. It is musculoskeletal, therapy through Back Center Clinics (physical therapists, orthopedists, psychiatrists) is usually helpful. Gait training and exercises, as well as support devices such as canes or crutches, may be helpful.*

*Only a very small proportion of patients with CMT have shoulder weakness. It is more common in the spinal muscular atrophies and dystrophies. It also may be musculoskeletal, and physical therapy may be beneficial.*

*Few patients with CMT have scoliosis or kyphosis as part of the disorder, while in other patients curvature of the spine is secondary to other factors.*

*In CMT the peripheral nerves are affected primarily, and the muscles are*

(cont'd on page 12)

## Letters to the Editor



*Dear CMTA,*

I am a recently diagnosed Type I and the only one in my family with symptoms. To know which of my relatives are at risk, it would be useful to learn from which of my parents I inherited the mutant gene, or if it arose *de novo*. But, because my parents are deceased, I have to rely on subtle clues, and am hoping you can help me with these subtleties.

My son is quite pigeon-toed, and this suggests that my mutant gene is autosomal dominant. My son has not yet been evaluated for CMT and it will be a while before I tell him he is at risk. Neither of my parents was pigeon-toed and I don't know if either had *pes cavus*. But a clue is that I have ridiculously wide feet (quintuple E), and so did my dad. My neurologist told me that many of his Type I patients have very wide feet, and he believes that this is of diagnostic value. However, I don't find the evidence compelling because his patient population is probably nonrepresentative and may be genetically homogeneous. I have found nothing in the literature on wide feet and Type I disease.

If I learned that large numbers of Type I patients across the country have very wide feet, I would find that evidence persuasive. It would not be helpful in this context to hear from individual patients with wide feet, but what would be very helpful is evidence, pro or con, that a large number have wide feet. Do wide-shoe sales people show up, salivating, at CMTA conferences? CMT patients, please write to me if you have useful information, and I will pass it on for a future issue of *The CMTA Report*.

I hope you will also write if you can help me find relief for one of my symptoms. Like many of you, I have muscle pain in my arms and hands, legs and feet. But the symptom that limits my activities to the greatest degree is muscle pain in my upper back, often between my shoulder blades.

Several clinicians told me that Type I related muscle pain occurs only in the distal extremities. They were unaware that at least four investigators independently found an association between scoliosis and Type I disease. I would therefore be very grateful to hear from Type I patients with symptoms like mine. Have any of you found relief through wearing a back brace? Have you had successful experience in this context

with a physiatrist or orthopedist in the New York City area?

Here's my address:

Mel Schechter  
382 Central Park West  
Apt 17F  
New York, NY 10025-6037

*Dear CMTA,*

I am a lawyer in South Central Pennsylvania. I was diagnosed with CMT in 1963. I had free follow up care through the Columbia Presbyterian Medical Center in New York through 1977 until the Federal funding evaporated. Through college, grad school and law school, I kept fairly active physically, although any kind of jumping or fast and sustained running was out of the question. To date, I have not had any upper extremity involvement whatsoever.

Approximately eight years ago, a physiatrist prescribed for me a molded ankle-foot orthotic support. The prescription was fulfilled by Don Zielke of Zielke Orthotics and Prosthetics, Inc., 1603 Rodney Rd, York, PA 17404. While distance running is now behind me, and being able to dunk a basketball is no longer so important, the MAFOs have permitted me a degree of physical activity I never thought possible. I play racquetball at least five days a week and, other than an occasional shattering of the MAFOs, I have had no problem whatsoever. The MAFOs sit in the shoe and run up the back of the leg over the calf. If there is a downside to using it, it is the heat that is generated by having the foot sit in plastic, particularly during the humid summer months in Pennsylvania.

Many policies of insurance cover MAFOs under the major medical provisions.

The old style polio braces of the past are long gone. They have been replaced by high tech, co-polymer plastics which are both lightweight and virtually indestructible.

Very truly yours,

J.K. York, PA

*Dear CMTA,*

Thank you for all your helpful answers to endless questions about CMT, the As-

sociation, support groups, physical therapy, and the like. If we'd known about you five years ago when my daughter and I were diagnosed, things might have gone a little easier. Our neurologist and orthopedic surgeon were both uninformed of CMTA's existence at that time, though luckily they were aware of the disease itself.

Thank you for working so hard to get the word out about CMT. We found you last fall through a magazine advertisement that was noticed by a special education teacher familiar with our daughter's case. What luck!

D.C. Michigan

*Dear CMTA,*

Thank you so much for sending us copies of your superb educational materials so promptly in response to our recent request for educational materials for our reference library. We appreciate your attention and are very grateful for your kind and generous gesture in gifting these to us!

We try to provide the best care possible for our patients. One of our priorities is patient education, since well informed patients take better care of their own health.

I do hope you will continue to send us your excellent *The CMTA Report* newsletter for our library - we look forward to receiving this regularly in the future!

I thank you once again on behalf of all our patients, and we look forward to utilizing your reference materials and to making good use of your help and guidance in order to improve the care and services we provide to our patients.

May God bless you!

Thanking you for all your kindness,

Dr. A.N. Malpani, Medical Director  
Community Health Research  
Programme, Bombay, India

*Dear CMTA,*

Last summer, my husband and I flew to Rhode Island to visit my brother and sister-in-law. While there, I found out that my brother had been diagnosed with CMT and he thought that I had it.

(continued on next page)

At first I was angry. I have four teeth that I take out to brush, a breast prosthesis, a metal hip, glasses, and a hearing aid. I didn't think I needed anything more. Already, my husband has said that when I die he isn't going to bury me, but he's going to take me to a flea market and sell my spare parts.

At first, I was hurt, but now I just think it's funny. A long time ago, I had accepted Christ as my savior and I won't be here anyway. Where I'm going, I sure won't need all that stuff.

Actually, I am happy to find out what is the matter with me. No more searching to find out if there is a relationship between my hearing loss and my clutziness. So, I have learned to cope. I watch where I put my feet, don't stand up or turn suddenly, etc. Actually, being in such a mad dash is one of my worst problems.

A couple of years ago, I stepped off the step stool the wrong way. I fell onto a cement porch and rolled down a couple of steps. I sat up and looked at my gory wrist and said, "Ann Landers says if it ain't broke don't fix it." It wasn't broken, but I thought it needed fixing. I knew I was perfectly capable of driving myself to urgent care, but I also knew that all of the family would be mad at me. My husband was out fishing so I called a daughter who lives nearby. That careless step cost both of us to miss church for a Sunday at the urgent care center and left me with two ugly scars on my already scared wrist.

I am 70 years old and have enjoyed life. So, I really don't mind it for me. But I'm not crazy about passing it on to our children.

My husband took our nearly six foot grandson fishing. Then he complained about how clumsy he was in the boat. I said, "Listen, when you get someone as big as he is and as clumsy as I am combined in one person in the boat, you have to expect problems."

Then, there are our daughters who can sprain an ankle falling over a shadow on the tennis court. And what about that beautiful little granddaughter who danced around on tiptoes as a toddler? I don't like that.

So, no more clawing my way up a waterfall to see if there might be a swimming pool up there. Or scrambling around the hills looking for a fern or another type of flower. And, ladders are a definite no-no for me.

Sure, there are quite a number of things that I can no longer do. But there are also a lot more things that I can do and I praise the Lord for every one of them.

Still it would be nice if all the containers that say "open here" would.

Thanks for a great magazine that is most helpful to all of us who have CMT.

Sincerely,  
D.H.B., CA

*Dear CMTA,*

I read the "workout" letters from other people with CMT with great interest. I have another point of view. Previously, before CMT, I was always very physically active in sports and other activities, but I gradually became unable to do those sports and activities. Over the course of about 8 years, even before I was diagnosed, I began to get overuse injuries and muscle strains from the same activities I had been doing all along. I started out "in shape," so, for me, there wasn't much I could do to stop the progression. As the disease progressed, I lost my ability to run, racewalk, bike and practice karate. I had been regularly lifting weights, too, for 4 years, under the supervision of a physical therapist. It became difficult to even get to the gym because of numbness and fatigue, and my legs refusing to move forward easily. Out of sheer determination, I persisted and went anyway, but I would get numb arms (not just forearms and hands) and shoulders, numb hips, legs, and feet, and blurry vision whenever I worked out. It was very difficult to get home on public transportation after a workout. Finally, I realized that I was not helping myself by working out, and I stopped going to the gym.

I continued to walk in the mornings, but I had to cut my distance gradually from 4 miles to 1 mile and choose flatter routes. When I began to need a cane, I continued my walks, but I did them less often. Finally, after 6 months with a cane, I had to stop exercise-walking completely because I was having more problems with being able to walk anywhere, with pain, and also with fatigue. Now I swim sometimes.

As a person with CMT, Type 5, not just my lower extremities are affected, but also my hips and shoulders and my balance, too. Sometimes working out and

exercising are not the answers to dealing with a progressive neurological disease.

I would encourage anyone who can work out to do so, but if the disease has progressed beyond being helped by working out, the person with CMT should alternatively try to be active in daily living and not overdo.

J.R., NY

*Dear CMTA,*

This letter comes to you from the Netherlands. I have been reading your publication since I visited a kind podiatrist to have some surgery on my toe. He immediately recognized my condition and told me about your association. I continue to count this man as a blessing to me because he put me on the track to better educate myself on my disease.

Rather than elaborate on how I discovered my disease, etc. I will just tell you this. I am a 25 year-old graduate student in a European MBA program who has a severe case of CMT but has learned to push onward and pursue my dreams with my faith in God and my faith in myself. I exercise in the gym 3 days a week, and have pursued biking as my manner of excelling athletically. A new "interest" here never called me again after I took him on a 2 hour bike ride and outbiked him. Little did he know that this was a personal victory I will never forget, because it proved to me that disease or no disease, there will always be things that I can pursue to find fulfillment. You are your own limitation.

But, on a practical note, this is for all the ladies with CMT who are frustrated about being invited to a formal dance and not being able to wear normal shoes with your dress. I twisted each of my ankles at least 8 times before I was ever diagnosed with CMT. So, I watched closely how trainers used to tape my ankles. When I go dancing, or out in formal wear, I tape my ankles and do great for the evening along with a pair of flat shoes that lace up half way. I can't walk very fast, or very far, but I can dance, and feel good about myself. Also, when I work out at the gym, I use lace-up ankle wraps that athletes with weak ankles typically wear. They cost about \$25 each and you can buy them at any athletic store. When I am either working with free-weights or biking, I find that I work more of my leg

(continued on p.13)



## Two CMT Conferences - Philly and New Orleans

The CMTA's East Coast CMT conference will be Saturday, September 10, 1994, at the Children's Hospital of Philadelphia (CHOP). CHOP is the children's facility of the University of Pennsylvania Medical Center. The conference is being sponsored by Dr. Philip Chance of CHOP in cooperation with the CMTA. The conference features Dr. Chance, a pediatric neurologist, speaking on "Current Genetic Research for CMT", and Dr. John Sladky speaking on "The Neurology of CMT". In the afternoon Dr. Carol Oatis, the physical therapist that many members know from our physical therapy VCR tape, will speak on "The Value of Physical Therapy for CMT Pa-

tients". Other speakers will discuss orthopaedic considerations for CMT patients. All of the speakers will allow time following their presentations for questions and answers. At the conclusion of the formal program, Ann Beyer, a doctoral candidate at Columbia University, will facilitate a workshop entitled "Taking Charge of Your Disorder".

Lunch is included in the \$20.00 registration fee. For attendees coming from out of town, via AMTRAK, the train station is six blocks from CHOP.

The CMTA is arranging a dinner following the conference for those who would like to stay and socialize and be enter-

tained by the Mixed Nuts, a professional comedy troupe. One of the members of the Mixed Nuts is Rex Morgan, the former Delaware Valley support group leader and a member of the Board of Directors of the CMTA. The total cost of dinner is \$23.00 per person. Dinner will be served in Penn Towers which is located directly across the street from CHOP. Penn Towers also offers rooms for conference attendees at a special rate of \$90 per room. Room registrations must be made by individuals and the special rate is available by mentioning the CMT conference at CHOP. The phone number of the hotel is 1-800-356-PENN.

### CMT Conference Registration

Name(s) of attendees \_\_\_\_\_

Address \_\_\_\_\_

Telephone Number \_\_\_\_\_

We are attending the Philadelphia conference.

\_\_\_\_\_ persons will be attending the conference  
@ \$20 per person \$ \_\_\_\_\_

\_\_\_\_\_ persons will be at the Saturday night dinner  
@ \$23 per person \$ \_\_\_\_\_

Total enclosed : \$ \_\_\_\_\_

We are attending the New Orleans conference.

\_\_\_\_\_ persons will be attending this conference  
@ \$20 per person \$ \_\_\_\_\_ enclosed

Please make all checks payable to the CMTA.

#### Registrations

for Philadelphia should be in the office by Tuesday, September 6th.

for New Orleans should reach the office by Monday, November 14th.

Return this form to:

Pat Dreibelbis - CMTA  
601 Upland Ave.  
Upland, PA 19015

Questions???? Call Pat at 1-610-499-7486.

### New Orleans

On November 19, 1994, the CMTA, in cooperation with Dr. Carlos Garcia, is hosting a CMT conference at Louisiana State University Medical Center. In addition to Dr. Garcia's presentation on "The Features of CMT", Dr. James Lupski, noted CMT researcher from Baylor Medical Center, will speak on "CMT and Genetic Research". This promises to be a very exciting CMT conference. Other presenters include Dr. Robert Dehne who will discuss orthopedic considerations, a physical therapist and an orthotist. As always, the speakers will leave time for questions and answers. Lunch is included in the \$20 registration fee. LSU has arrangements with the Clarion Hotel to offer attendees a room for \$60 per night. Again, each individual must call the hotel for reservations and state that they are attending the CMT conference at LSU. The phone number is 1-800-824-3359. Ask for Mike DiLeo.

The CMTA strongly urges you to attend either (or both) of these conferences. The information you will receive is invaluable. Moreover, the socializing with other CMT patients, parents, and family members is equally invaluable. We hope to see you there! §

#### Ask the Doctor - cont'd from p. 9.

*affected only secondary to the nerve involvement. The longest nerves are initially affected, and that is why the weakness starts in the feet and legs, then the hands, but rarely involves muscles clinically in the back.*

*Most patients with CMT can remain active throughout life, even through the eighth decade. §*

muscles wearing these lace-up wraps than if I wear my AFO's. The rest of the time, however, I wear my AFO's. These tips have helped me better manage my disease and allow me to enjoy my age to the fullest.

Good Luck and don't give up hope.  
Carpe diem!

G.R. The Netherlands

*Dear CMTA,*

I read every newsletter that comes to me and I have trouble understanding how people can be so down on themselves and think that they are no good. I was diagnosed with CMT when I was 5 years old, and I have had my heels lowered, my hammer toes fixed, my ankles fused, my tendons moved, and I have never felt better in my life. There is nothing stopping me now. I read some of these life stories and I can't believe that their lives have been so unhappy. That's depressing. Here is a neat twist on CMT:

I'm not the last picked in Gym.

I'm involved in many Varsity sports at my high school.

I'm very self-confident in everything I do.

I'm very popular and a Junior Class officer.

I'm looked at as "Big B-Loy" and not the kid who walks funny.

I feel that everyone with CMT to whatever degree has something special to offer to the world. I'm one of the lucky CMT patients who don't have it so bad. I see all the other kids in the duPont Institute. I saw one boy with something with metal rods sticking out of his thigh strapped to this cage. I say to myself, "I just walk funny."

The two most horrible things that I remember was, first, when I was about 10 years old. The cast man at CHOP came in with a drill and said, "Ready for those

pins in your hammer toes to come out?" I remember screaming at the top of my lungs, I was so scared. The second time was when I was 14. I was sitting in the waiting room waiting to get my first cast changed. When I went into the curtained off section and laid on that table and they started cutting it, it felt like I was getting my leg cut off. People were staring at me because I was crying, cursing, and telling my Mom through teary eyes to make them stop.

If you are getting surgery soon, don't be alarmed. Everything will be okay.

B.L. Blue Bell, PA

*Dear CMTA,*

I appreciate all the correspondence I have received from the CMT Association. It is comforting to hear from others and know that I'm not alone in dealing with CMT.

When I was 15 years old, I had to wear a back brace for one year because of scoliosis, this was just the beginning. Slowly, my coordination got increasingly worse. The doctors kept saying I was growing fast and it was affecting my coordination.

My mother approached the medical doctor about the fact that I wasn't able to keep my balance and couldn't pick up my feet when I walked. She mentioned the fact that stroke victims learn to walk again with therapy. Why wasn't there any help for me?

I was sent to a doctor of medicine and rehabilitation (physiatrist). One look at my feet and the doctor was 90% sure what was wrong. He ran the test on my muscles and nerves. With that, he diagnosed it as being CMT. He then prescribed braces, physical therapy, and occupational therapy.

I was very depressed because I wore a very, very uncomfortable back brace for one year so I was not receptive to the braces at all.

What a wonderful Christmas present at 16 years old. I could walk again, almost normally. Because of the braces, I can now drive, play basketball, racquet ball and keep up with my friends pretty well. Because of keeping active, the braces have to be replaced, one every 6 or 7 months. I feel it is worth it though because I am able to keep active.

Even though having CMT is frustrating sometimes, I try to keep a positive attitude and look for all of the good things that my life has been blessed with. I am now attending Pueblo Community College, planning on majoring in youth ministry. The following is an experience that I had with my church youth group.

"Having CMT is no big deal to me most of the time, except for wearing braces and not being able to feel much from my knees down. Usually, people meet me and like me before they meet my CMT. However, one experience I had at the age of eighteen was one that I will never forget. My church youth group of twenty kids went to Water World, which is miles of concrete, water, and babes. I can remember my friends walking slowly with me because I couldn't wear my braces. That made me self conscious enough, but that was a drip compared to the flood that was about to come. After we rode a slide, I noticed that we were at the bottom of a huge hill. On the walk up my "uniqueness" became more apparent as my right foot started to hurt more and more with every step. This made my gait so uncoordinated that I had to stop. I looked back and at each place my foot had touched was pool of blood. What hurt even more than my foot was the humiliation of being carried up the hill with all eyes on me. Even though this experience was unforgettable, I choose to look at my CMT not as something that makes me different, but as something positive that makes me a unique person."

D.M. Pueblo, CO

*Dear CMTA,*

I enjoyed reading the June 1994 CMTA Report. Thank you! The report on page 2 entitled "Update on Nutrition Supplements" contains a very common error in fact.

Quote: "the generic drug industry is required to abide by very stringent government standards in order to prove that their drugs are absolutely bioequivalent to brand name drugs." Approximately Ten (10) percent of the generic drugs now available are not equivalent! Also, instead of running your own dissolution tests, the consumer can depend on the USP designation or the reputation of the manufacturer. The same caveat applies to prescription drugs!

E. A. Registered Pharmacist

Write us:



Letters - CMTA

601 Upland Ave.  
Upland, PA 19015  
Ph. 610/499-7486

# CMTA Contacts

Following is a list of CMTA contact persons and support group leaders. There are many CMTA support groups, but more groups are needed. The CMTA will help you set up a group in your area. For information about forming a group or being a local contact person please inform the CMTA by mail or call 610-499-7486.

\*denotes support group leader

## Alabama / Greater Tennessee Valley

\*Bill Porter 205/386-6579W  
205/767-4181

## California

\*Janice Hagadorn 805/985-7332 after 5  
(Oxnard/Thousand Oaks)  
\*Sheila Levitch 805/254-5322  
\*Denise Miller 805/251-44537  
(Canyon Country/Saugus)  
\*Gary Oleze 619/944-0550  
10am-5:30 pm 619/436-2116  
(San Diego)  
\*Freda K. Brown 707/573-0181  
(Santa Rosa)  
Eda Adams, will return calls  
916/677-6460  
Jeanne Amour 408/258-9122  
Clair Bumgarner 209/874-4963  
Felice Gail Viggers, 805/492-2840  
Verna M. Sabo, 818/892-6706  
Mary Micalizzi, after 6pm  
619/441-2432  
Bob Hedge, 9am-5pm 310/645-2761

## Colorado:

\*Dr. Gregory Stilwell 719/594-9920  
(Denver area)  
Roberta Cummings, 719/846-5611

## District Of Columbia:

\*Lorraine Middleton, 6pm-9pm  
202/362-4617

## Florida:

William Brady 904/443-6271  
Mary Beeler, 9am-8pm 407/295-6215  
Harold Wilson 407/465-3656  
Pat Ports, M, W, F, 4pm-9pm,  
407/965-3691  
Joe Ellenbogen 305/921-4660  
Edward Carhart, 9:30am-5:30pm  
305/567-1066  
Beatrice Bannister 407/737-3267  
\*Robyn Cohen 407/622-5829  
8pm-9:30pm M-F  
weekends anytime

## Georgia:

Nancy Lee McCutchen 404/925-1020

## Louisiana

Bobbie Marberry 504/872-0895

## Maryland

Jean Iler 410/987-5432  
Linda Ember Miller 410/882-4019  
Robert Kight 410/668-3054

## Massachusetts

Wayne Cardillo 413/298-3156  
\*Donald Hay 9am-7pm, 617/444-1627  
(Boston)  
Jim Lawrence 508/460-6928

## Michigan

Robert D. Allard 517/592-5351  
\*Suzanne Tarpinian 313/883-1123  
(Detroit)  
Laurie Vasquez 517/893-4125

## Mississippi

\*Julia Prevost 601/885-6482  
\*Henry & Brenda Herren 601/885-6503  
(Jackson)

## Minnesota

Grace Wangaard 612/496-0255

## Missouri

Allan Degenhardt 816/942-1817  
\*Ardith Fetterholf 816/763-2176  
voice mail 816/756-2020

## New Hampshire

Mary Nightly 603/598-5451

## New Jersey

\*Janet Saleh 908/281-6289  
(Somerville)  
\*Teresa Daino 201/934-6241  
(Englewood)  
Linda Muhlig 609/327-4392  
Gary Orson, Mon-Fri 6pm-10pm  
& weekends 609/584-9025  
Russell Weiss 908/536-6700

## New Mexico

Jesse Hostetler 505/536-2890

## New York

Joe Ehman 716-442-4123  
Internet:KOLOB@Multicom.Org  
Alan Latman 800/227-1343  
\*Diana Eline 201/861-0425 before 9pm  
(New York City)  
\*Abby Wakefield 212/722-8052 (NY)  
\*Lauren Ugell 516/433-5116  
(Long Island)  
\*Neale Bachmann /716 554-6644  
(Rochester)  
\*Bernice Roll 716/584-3585  
(Rochester)  
\*Kay Flynn 914/793-4710  
(Westchester County)  
Amy Gardner 518/272-6384  
Angela Piersimoni, after 2pm  
607/562-8823  
Sharon McAvey, afternoon & evening  
718/380-3792  
William Carrington, 4pm-11pm  
718/486-6953

## North Carolina

\*Susan Salzberg, 7am-3:30pm  
919/286-0411 x 6586  
5pm-9pm 919/967-3118  
(Durham)

## Ohio

Roger Emmons 216/286-6485  
Suzanne Lammi 513/339-4312  
\*Norma Markowitz 216/247-8785  
(Cleveland)

## Oklahoma

Leah Holden 405/255-4491

## Oregon

\*Mary Elizabeth York 503/246-4939  
(Portland)

## Pennsylvania

\*Dennis Devlin 215/269-2600 work  
610/566-1882 home  
(Delaware Valley)  
Patricia Zelenowski 717/457-7067  
Camille Walsh 215-747-5321  
Janet Fierst 412/487-0757  
Mary MacMinn 215/322-1073  
Carol Henderson 215/424-1176

## Rhode Island:

Robert Matteucci 401/647-9154 PM

## Texas

Dr. Karen Edelson, D.P.M.  
214/542-0048  
M,T,Th, 8:30am-5pm, 214/542-0122  
Tony Collette, 1pm-8pm, 713/699-8432  
Ken Kerby 817/282-9329

## Virginia

\*Mary Jane King 804/591-0516  
(Tidewater)  
\*Thelma Terry 804/838-3279  
(Tidewater)

## West Virginia

\*Joan Plant 304/636-7152 after 6pm  
(central)  
L. Ben Simmers 304/693-7731  
Beverly Simmers 304/364-5309  
Ronald & Rebecca Sampson,  
304/636-7449 24 hours  
Barbara Compton 24 hours  
304/636-5456

## Washington

Marlene Russell 206/484-3116



### Blood Test Available

The blood test for diagnosing CMT Type 1A found on chromosome 17 is available from Genica Pharmaceuticals. They can be reached by calling 1-800-394-4493, ext. 106. Ask for Sarah Quiry, customer service representative. A physician must order the shipping kit. The cost of the test is \$395.00. §

### Honoraria and Memorials

IN HONOR OF:

By:

Pearl Sand	M/M P. Becker Lorna & Arthur Carroll Robert & Phyllis Sand
Rebecca Sand	Ethel Vill M/M Richard Alexander Ellie Shaffer Samuel & Lillian Blender
Minna & Hart Wurzburg	Marge & Bill Sherman Mary & Floyd Laper & family
Marjorie Sachs	
Paul Isaacson	M/M Ron Catalan
Diane Higgins Kosik	M/M Joseph Higgins
Janet & Eric Bowden	Keith & Louise Petersen

IN MEMORY OF:

Adelaide Aldridge	by Wilma Leonard by Bud and Ann Oliver by Erma Covault by Jean Stafford by Ruth Polata
Mary Tarpinian Sophia Bradwick Virginia L. Reed James O'Neil Julie Held	by Sue Tarpinian by R. Gordon Bradwick by Mrs. W.O. Keneipp by Grace Wanggaard by James Held

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The opinions expressed in the newsletter are not necessarily those of the Charcot-Marie-Tooth Association. The material is presented for educational purposes only and is not meant to diagnose or prescribe. While there is no substitute for professional medical care for CMT disorders, these briefs offer current medical opinion that the reader may use to aid and supplement a doctor's treatment.

## CMTA Membership/Order Form

Name: \_\_\_\_\_

Address: \_\_\_\_\_

Phone Number: \_\_\_\_\_

Tell us about yourself:

- |   |   |
|---|---|
| <input type="checkbox"/> CMT Patient          | <input type="checkbox"/> Medical Professional |
| <input type="checkbox"/> Interested Supporter | <input type="checkbox"/> CMT Family Member    |

Enclosed is:

- |                                |                                |
|--------------------------------|--------------------------------|
| <input type="checkbox"/> \$25  | <input type="checkbox"/> \$50  |
| <input type="checkbox"/> \$100 | <input type="checkbox"/> other |

for my membership in the CMTA (newsletter included in membership)

- At this time I cannot contribute to the CMTA but would like to receive mailings.

### Publications and Tapes available from the CMTA

(Check to order)

- VCR Tape - CMT Neurology (\$15)
- VCR Tape - Physical Therapy & Occupational Therapy (\$15)
- VCR Tape - CMT Genetics (\$15)
- VCR Tape - Orthopedic Surgery & CMT (\$15)
- Handbook (16 pp.) - CMT FACTS I (\$3)
- Handbook (24 pp.) - CMT FACTS II (\$5)
- Transcript - San Francisco CMT Conference (\$5)
- Letter - to Medical Professionals regarding the drug list (free to members with self addressed stamped business envelope)
- List - Physician Referrals (by state) (please send SASE) please list states: \_\_\_\_\_
- CMT Informational Brochure (gray brochure) (one copy free with self addressed stamped business envelope)
- CMT Informational Brochure (gray brochure) in Spanish (one copy free with self addressed stamped business envelope)

Contributions are tax deductible.  
Please make checks payable to the CMTA.

Total amount enclosed: \_\_\_\_\_

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.

## MEDICAL ALERT



### ***Certain Drugs Toxic to the Peripheral Nervous System***

This is a list of neurotoxic drugs which could be harmful to the CMT patient.

Adriamycin  
Alcohol  
Amiodarone  
Chloramphenicol  
Cis-platinum  
Dapsone  
Diphenylhydantoin  
(Dilantin)  
Disulfiram (Antabuse)  
Glutethimide (Doriden)  
Gold  
Hydralazine (Apresoline)  
Isoniazid (INH)  
Mega Dose of Vitamin A  
Mega Dose of Vitamin D  
Metronidazole (Flagyl)  
Nitrofurantoin  
(Furadantin, Macrochantin)  
Nitrous Oxide  
(chronic repeated inhalation)  
Penicillin  
(Large IV doses only)  
Perhexiline (Pexid)  
Pyridoxine (Vitamin B6)  
Taxol  
Vincristine

Lithium, Misomidazole and Zolof  
can be used with caution

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

### ***Referrals Available***

The CMTA has compiled a list of neurologists, orthopedists, physiatrists (a physiatrist is a physician trained in physical medicine and rehabilitation) and podiatrists who have a special interest in CMT. We can also access respiratory specialists. Additionally, we have listings for pedorthists. A pedorthist is a practitioner who provides care to the patient by fitting orthopedic shoes and devices, at the direction of and in consultation with physicians.

To receive any of these referrals send a stamped self-addressed business-sized envelope indicating the geographic areas needed to: CMTA, 601 Upland Avenue, Upland, PA 19015.

For referrals for a hand surgeon contact the American Society for Surgery of the Hand, 6060 Greenwood Plaza Blvd., Suite 100, Englewood, CO 80111-4801, Ph.303/771-9236. §

## CMT...

- ..... is the most common inherited neuropathy, affecting approximately 125,000 Americans.
- ..... is also known as peroneal muscular atrophy and hereditary motor sensory neuropathy.
- ..... is slowly progressive, causing deterioration of peripheral nerves which 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) is sometimes present.
- ..... 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.
- ..... 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 the focus of significant genetic research, bringing us closer to answering the CMT enigma.
- ..... Type IA can now be diagnosed by a blood test.

## ***THE CMTA REPORT***

*information on Charcot-Marie-Tooth Disorders  
from the*

### **Charcot-Marie-Tooth Association**

Crozer Mills Enterprise Center  
601 Upland Avenue  
Upland, PA 19015

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