

# THE CMTA REPORT

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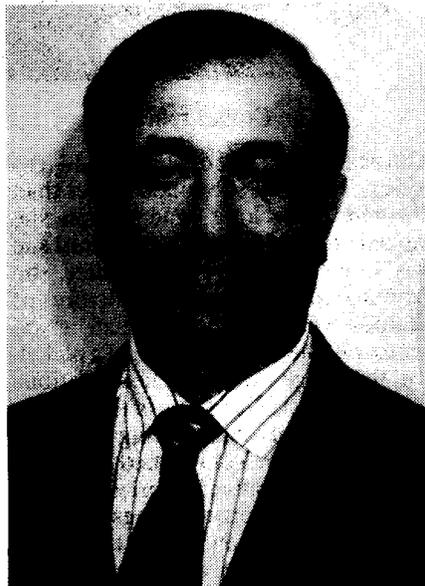
Providing information on Charcot-Marie-Tooth disease (a.k.a. Peroneal Muscular Atrophy or Hereditary Motor Sensory Neuropathy), the most common inherited neurological disorder. Contents © 1992, CMTA. All rights reserved.

## Charcot-Marie-Tooth Disease and Tremor

Francisco Cardoso, M.D.  
and Joseph Jankovic, M.D.

*Editor's note: The information for this article was obtained by the authors from a survey which appeared in the Winter 1991 issue of THE CMTA REPORT. We wish to thank all who participated in this survey.*

The primary goal of this research project was to study the relationship between Charcot-Marie-Tooth disease (CMTD) and tremor. In the first part of the study we examined 7 patients with CMTD referred to the Baylor College of Medicine's Parkinson's Disease Center and Movement Disorders Clinic. The following movement disorders were observed in these personally examined patients: 6 patients had a postural tremor, noticed when the patients were using their hands or holding their arms outstretched in front of the body. Hand tremor present at rest was noted in 3, and Parkinsonism (combination of stiffness of the muscles with slowness of the movements and tremor at rest) and dystonia (abnormal spasms



Dr. Jankovic of the muscles causing repetitive movements or abnormal postures) were present in 2. There was no relationship between weakness, loss of sensation, and the severity of tremor to suggest that the movement disorders were directly caused by the neuropathy, but were probably somehow linked to the CMT gene.

In the second phase of the study we surveyed members of the Charcot-Marie-Tooth Association (CMTA) in an attempt to determine how common various movement disorders are in CMTD. Over 2400 questionnaires were published in the CMTA newsletter. Of the 201 patients who responded to the survey, walking difficulty was reported by 87%, weakness 79%, and foot deformity by 71%. Tremor, present in 40% of the subjects, was usually first noted when the patients were 36 years old and mostly involved the hands. Family history of tremor was more frequent in the group of patients who complained of tremor. Medications such as propranolol (Inderal) and primidone (Mysoline) improved

tremor in most patients treated. In the last section of the questionnaire the subjects were asked to write "Today is a nice day" and to draw a spiral. The patients with a complaint of tremor had worse handwriting and drawing samples.

We concluded that movement disorders, particularly postural tremor, occur frequently in patients with CMTD. Essential tremor (ET) or benign familial tremor, the most common movement disorder, is a genetic condition characterized by the presence of postural tremor of the hands. Other characteristics of ET include tremor of the head, onset in the fourth decade of life, and improvement with propranolol and primidone. The overlap in clinical features between CMTD associated tremor and essential tremor (ET), the high frequency of family history of tremor, and the lack of a relationship between the severity of tremor and of the peripheral neuropathy suggest that the tremor in CMTD is probably related to ET. §

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### Upcoming CMT Conference

April 1993  
Wilmington, Delaware  
*details in the next issue*



*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.*

# CMT... A Personal Narrative

by Ivi Collier

(Editor's note: Ivi is the mother of current poster child, Sarah Collier)

Like so many others, I grew up without a diagnosis of CMT and with no accommodation for my CMT. This struggle has profoundly affected my life. I am determined that other children will have the support they need to be all that they can be in this world. As the parent of a child with severe CMT, I am convinced that our relationship is somehow key to her continuing adjustment to her ever-changing physical condition.

My father and most of his eight brothers are affected by CMT (which we vaguely referred to as the Nelson walk) in varying degrees. As I grew and saw their challenges and experienced my own, I found myself drawn to community service, wanting all children to have a fair shot at happy lives. This interest led me along a circuitous route to serving on the board of directors for the Maryland Center for Independent Living, and my eyes were really opened. I saw what it meant to live in America when you have a disability. I met many talented and capable people who were captives of the care system that was in place, or, worse, that was not in place. I had a friend who paid another person's attendant five dollars to feed him a piece of pie because his own attendant was off for the day. These experiences strengthened my resolve to somehow change the way that people with disabilities were kept from participating in society. It seems ironic now to think that a few years later I would have a child with a disability that would severely threaten her chance to have all the opportunities that she deserves.

When Sarah was born in 1982, I'm embarrassed to say that I really knew nothing about CMT, least of all what it was called. When she was pronounced fit and fine at birth, I breathed a sigh of relief and felt that she, like her older sister, had escaped having to face the physical difficulties that I have. I happily took my healthy baby home to begin our new family life. As time went on, Sarah grew and prospered, but occasional concerns popped up. Once, when she was about 8 months old and napping in her crib, I commented to my husband about the way she held her hands. I remembered that when I was very little and my grandfather slept in his chair on the porch he let his hands sit on his legs just like she was doing.



The Collier family

Although she talked clearly and co-gently very early, she rolled rather than crawled for a long time. When she walked, she held on to things until she was 16 months old. As a toddler, she drew accurate and detailed pictures of bunnies when the pediatrician asked her to, but they were the size of a nickel, not the large, vague generalizations normally seen. The doctors that I consulted were fairly patronizing and went on about how accomplished she was, making me feel silly for worrying about *how* she did the many things she had learned, rather than the fact that she could do them. She was beautiful and happy and quite healthy and I was beginning to be looked at as an over-protective mother, too involved with her child. It was my interest in disabilities that finally tipped the scales. At the pediatrician's office one day, she was drawing a picture. By now, she was nearly five years old. As she reached for the pen, I said, "Look, see how flat her hand is? The way she holds her hands reminds me of the way a quadriplegic would approach picking something up." The doctor asked what I knew about quadriplegia and in the ensuing dialogue, I learned that he had a daughter who had had a brain-stem stroke, and was nearly completely paralyzed. Suddenly, it seemed I had validity and he gave me a great referral. Sarah was diagnosed with CMT within a few weeks. Coincidentally, my father was diagnosed by another doctor the same week. I was diagnosed by inference. (Editor's note: *Diagnosis of the child first is a story we frequently hear.*)

Since that time, CMT and its implications have been an ongoing aspect of our life. Because we have a large, yet close-knit, extended family, we were

very interesting to researchers, and many of us have taken part in studies. Seeing how seriously Sarah's CMT affected her really brought my cousins, uncles, and others to go to bat for her. Many of them had to push themselves to be studied and have the tests done, but they all came through for her. It means a great deal to me and helped me deal with the loss of the "ideal" that so many of us must endure.

For myself, I was suddenly a sword. When Sarah began school, we saw firsthand the prejudice and ignorance that is still so prevalent. Although her home school is completely accessible, school personnel wanted to know when she would go to a special school. I realized that if Sarah were to lead a normal school life, I needed to learn a lot about accommodation as well as how to educate the professionals with whom I would be dealing. Through the CMTA, the MDA, and other resources I've found around the country, I learned how Sarah could be successful in school. She is a "Gifted and Talented" student and is happily accepted by classmates. However, it remains an ongoing process to have her school program adjusted.

Last year she was not included in an advanced placement math class because it met in a trailer and Sarah sometimes needs to use her wheelchair. It took some effort to get this cleared up, but she was included. We are able to head off some potential problems by meeting with the school and all the teachers before the start of a new year. They get a chance to meet her (and me) and to get an idea of how very capable she is. When one administrator asked, "how will we know what she has learned if she can't write it down?" My husband replied, "You could ask her!" We were asked, "Would you prefer we call her handicapped or disabled?" We replied, "We prefer you call her Sarah." Often the solution is very easy. The hard part is getting people to see that the way they have always done things isn't the only way. As Sarah says, by Christmas each year the teachers have got it and things go pretty smoothly.

My work for Sarah has led me to a new career. I am the coordinator of a Parent Resource Center that helps other parents understand their options and rights under the law. This sounds rather

(continued on page 4)

## Items of Interest



**ITEM 1:** Gerad Schuldies, grandson of member Norma Thomson, has been named poster child for the Greater South Dakota Area Chapter of the MDA. He was diagnosed with CMT at the age of 6. Gerad is also legally blind and because he is adopted, no family history of his disorder is available. Norma writes that Gerad is now 10, a very happy active little boy who doesn't know that he falls and gets hurt more than most kids his age. She calls him a "blessing" to his family.

**ITEM 2:** Some Tips On Talking With Your Doctor. (Used with permission of Health Ink Publishing Group.)

1. Be assertive. Make sure you get across everything that brought you to the doctor's office.
2. Don't be shy and don't be intimidated, either.
3. Make a list, on paper or in your head. Before you go, focus your worries by talking to a friend or family member about them.
4. State your most serious complaint first, as clearly as you can.
5. Ask these questions: Where is the problem? What caused the problem? How serious is it? What can be done about it?
6. Tell your doctor what you want from him/her, and ask him/her to tell you what he/she wants you to do.
7. Don't take things personally. If the doctor interrupts you, it's probably because he or she is behind schedule and feeling the time pressure.

Some Other Things To Keep In Mind:

1. Be punctual. If you must cancel an appointment, do it as soon as possible.
2. If it is a first visit, ask what information you should bring with you, such as medical records from other physicians or x-rays. Bring information on your health insurance.
3. Bone up on your family history. Since CMT runs in families, your parents, grandparents and great-grandparents could be relevant.

4. Be informed. Read up on health issues. Broaden your knowledge, but don't make your own diagnosis.

**ITEM 3:** The Sierra Club has published a book called Easy Access to National Parks as a guide to people with disabilities. The book was written by Wendy Roth who has Multiple Sclerosis and Michael Tompane, a strong and athletic individual. The two of them visited 41 national parks, and their descriptions of the sights and the accessibility are both practical and lyrical. This is a most readable book and one which inspires the reader to plan a visit to a national "treasure" as soon as possible. The philosophy which governs this book is that access is for everyone and that the challenge of the wilderness is amazingly equalizing. The book can be obtained at bookstores or by direct mail from: Sierra Club Store Orders, 730 Polk St, San Francisco, CA 94109. \$16.00 plus \$3.00 for postage and handling.

**ITEM 4:** Enabling Romance: A Guide to Love, Sex, and Relationships for the Disabled (and the people who care about them) by Erica Klein and Ken Kroll (Harmony Books, 1992, \$22.50) is a comforting and informative guide to romantic relationships for the "forgotten minority" - those Americans with disabilities.

**ITEM 5:** There has been a new drug added to the neurotoxic drug list. Please add this to any card or paper that you now use. New lists will be published later. The drug is perhexiline (Pexid) and is prescribed for angina pectoris.

**ITEM 6:** ALERT! \$\$\$\$\$ The time is coming for the annual United Way and Combined Federal Campaigns (CFC). If you are an employee of the Federal Government, our number in the CFC is 1255. You can designate part or all of your contribution to the CMTA which is a member of the Independent Charities of America. If your place of business solicits for the United Way Campaign, you can designate your pledge to the CMTA (designated donor numbers vary by campaign areas.) Please remember to request that your name be released to us if you wish to receive a thank you and have your do-

nation records updated. The amount of the gift need not be revealed to us, only the fact that you have directed your pledge to our organization.

**ITEM 7:** Cephalon, a new biotechnical company, has announced a research and development partnership to focus on the development of products to treat neurodegenerative diseases. Among the disorders that Cephalon plans to concentrate its research programs on are ALS, Alzheimer's disease, spinal cord injuries and Charcot-Marie-Tooth disease. Cephalon has demonstrated in preclinical trials that Myotrophin, a recombinant form of human insulin-like growth factor-1, promotes the survival of neurons. They hope to develop Myotrophin to treat the listed disease conditions. Please do not call this office or Cephalon because the work is in its very early stages. We will keep our readership updated as more information becomes available.

**ITEM 8:** CMT scientists will be meeting at the Human Genetics Meetings in San Francisco in November to discuss their current genetic research. All members of the Medical Advisory Board of the CMTA who are concerned with genetic research, as well as Chairman of the Board, Dr. Robert Lovelace, have been invited to this meeting. We will report on the meeting in the winter issue of the newsletter.

**ITEM 9:** At the Human Genetics Meetings, there will be a ten poster symposium exclusively on CMT disease. This is the first time this has occurred at a major medical meeting and will represent the work of 10 major CMT research projects.

**ITEM 10:** The Alliance of Genetic Support Groups announces that the Health Insurance Resource Guide is now available. Developed with a grant from the March of Dimes Birth Defects Foundation, the Guide helps provide an understanding of the health insurance system in the United States. It also gives specific information about what people need to know in order to find help. It is available for \$10 from "The March of Dimes" Supply Department, 1275 Mamaroneck Ave, White Plains, NY 10605. Computer # 32-577-00.

This year has been another year of growth and productivity for the CMTA. Our CMT patient base is continually increasing as the CMTA becomes known to the CMT community and the medical community. New publications this year include the transcript from the San Francisco conference, the restaurant alert card and the neurotoxic drug alert card. Publications that are in production are the Physician's Handbook and CMT FACTS II. We anticipate CMT FACTS II will be available in early 1993, and the Physician's Handbook will be published later in 1993. Both publications will be announced in *The CMTA Report*.

Officers elected at the annual meeting of the Board of Directors are Karol Hitt, president; J. Rodman Steele, Jr., vice-president; Diane Freaney, treasurer; Donald J. Perrella, corresponding secretary; and Robert Daino, recording secretary. Other Board members are Rex Morgan, Jr., Robert E. Lovelace, George Crohn, Jr., Ann Lee Beyer, Diana Eline, and Gary Griffith. The CMTA's financial position is stable and sound. Our employee Patricia Dreibelbis became full-time in May. The increase in Pat's time was necessary due to the growth of the CMTA and possible because of your financial support. If you would like a copy of the CMTA's annual financial report, write to the office and request it.

Miami, FL and Chicago, IL were the sites for 1992's CMT patient/family conferences. Dr. Walter Bradley, Neurology Chairman of the University of Miami Medical Center, and Dr. Barry Arnason, Neurology Chairman of the University of Chicago Medical Center were our hosts. Again, we were able to educate and provide support to many, many CMT patients and families. In May we attended the American Academy of Neurology (AAN) meetings and in October the American Neurology Association (ANA) meetings.

At these meetings the CMTA's Medical Advisory Board (MAB) met and discussed current CMT research and therapies. Dr. Robert E. Lovelace, Columbia University College of Physicians and Surgeons, is Chairman of the MAB, and Dr. Roger Lebo, University of California San Francisco, is the research liaison. Dr. Gareth Parry, University of Minnesota, is the Editor of the forthcoming Physician's Handbook. This publication is being written

for use by primary care physicians (pediatricians, family physicians, and internists).

Sarah Collier, of Baltimore, MD, was chosen to be our 1992 Poster Child. Sarah and her mother Ivi are active in programs for the disabled, and are wonderful ambassadors for the CMT community. Sarah is the 1992 recipient of the "Yes I Can" award.

A major change this year occurred in the VCR tape program. We went from a rental program to a sale program. This change was necessary because of the demand for the tapes and all of the problems inherent in a rental program. The other major change was the purchase of a new computer for the office. This has been a wonderful addition to the office and a major work enhancer. The speed of this new computer amazes and pleases us greatly.

We offer our congratulations to Mary Beeler and Walter Kelly Jr. for sharing the Rebecca Sand Volunteer of the Year award. These people are only two of the many volunteers who give their time and talents to the CMT community. We offer our thanks and congratulations to all of you who volunteer your time and talent so freely and generously. You make the programs of the CMTA possible. At this time we are mailing this newsletter, *The CMTA Report*, to about 4,500 persons in the United States and in 30 foreign countries. This number is continually growing, and this growth is a reflection of the increasing awareness of the CMTA and its programs. If each of you sent the name of a CMT patient (or family member) currently unknown to us, then obviously, our roles would immediately double. This is something you could do right now, and we urge you to do so.

The CMTA is in existence because of a great need and because all of us work together. The Board and I thank you for another successful year of growth and service. §

*Karol B. Hitt, President*

### **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.

more grand than it is - I am the only staff person and do everything myself. I work with parents individually and also offer a course on how they can become partners in their child's education. I go to Sarah's class the first week of school each year and do some activities that help the children understand Sarah's challenges. She and I also go to schools around the state as part of the DISABILITY Awareness Project. We talk about the accommodations we need and about the fun things that Sarah does. She's won medals for swimming, track and field, and horseback riding. She's played the Dormouse in the Community Theater, and has even made a commercial

I am committed to inclusion for everyone and believe that the key to this lies with educating and helping the children. If we all understand the power that we have and exercise the skills we've developed, the world will be a better place, not just for those with disabilities, but for all people. A former candidate of national office said that it was better not to waste money on "special education" but to concentrate efforts and dollars on the best and the brightest. This not only ignores the fact that those with disabilities often are the best and the brightest, but the fact that people with disabilities are going to take their rightful place in society. By equipping them as children, we not only minimize the stigma that goes with being different, but we save on the expense involved in helping adults become full-fledged citizens.

I will always regret that my child has CMT, just as I regret that any child will have a harder life journey. There is an agreement between myself and my family members that when I die they will enclose a pair of 4 inch red spiked heels in my coffin in the hope that I will not need "sensible" shoes wherever I go. In the meantime, I look at my daughter and see the opportunities as well as the obstacles. My life is better for the friends I've met and made along the way. Knowing and loving successful adults with disabilities keeps my spirit and courage high. In the process of adjusting and growing and living, I have learned things I never would have otherwise. I still have far to go and much to learn. I look forward to it. §



## Letters to the Editor

### Dear Editor,

A recent article was published by you having to do with a patient on whom I operated using the Ilizarov technique for correcting and maintaining correction of severe deformities of the foot and ankle. Dr. John Hsu, a consultant for the CMTA, made several comments. Clearly, Dr. Hsu's comments are based on his own experience with patients with CMT in regard to this particular procedure.

Obviously, patients who have had severe deformities due to Charcot-Marie-Tooth disease have two problems. First is abnormal muscle balance and second is the boney deformities associated with long-standing malposition of the foot.

Traditional operations to correct boney deformities require that the foot be shortened. The more severe the deformity the shorter the foot would become. This is a great problem since shortening the foot too much would lessen the base for standing.

In such instances, the types of more conventional surgeries that were described by Dr. Hsu have problems for which the Ilizarov technique is of enormous benefit. In using this technique, we are fusing the bones together, lengthening bone, tendons (muscles), blood vessels, nerves and skin. Although there is a significant amount of activity that is required on the part of the patient during the process of correcting the foot, the actual length of time that the patient is under treatment is perhaps half again as long as the conventional methods. For example, instead of the healing process taking two to three months, it may take three and a half to four months.

The nature of the deformities that my patient, Mr. Enowitz, had were very severe. If he had had more conventional surgery, his feet would have been shortened significantly. At this time, his feet have an essentially normal configuration and a proportionate size to the rest of his leg.

I would agree with Dr. Hsu that this is a method which requires special levels of attention and knowledge and should be entertained in specific instances. It is not necessarily appropriate or good for every patient with a foot deformity.

However, when a patient has problems of this dimension, the physicians or clinics that he goes to should have this type of treatment as an option.

Sincerely,

Alfred D. Grant, M.D., F.A.C.S.  
Hospital for Joint Diseases  
Orthopaedic Institute  
301 East 17th Street  
New York, NY 10003

### Dear Editor,

Thank you for publishing your interesting article regarding orthotics and the orthotist. I found your article to be thorough and informative. I was disappointed to note the absence of the mention of podiatric and orthopedic foot surgeons and their role in orthotic management. Foot and ankle orthoses have been greatly refined and improved in part thanks to the contributions of physicians in these fields. Many such physicians actually perform casting and fabrication of these devices as well as the prescription of them. In our community we work closely with the orthotist to prescribe, cast for and fabricate the best possible device for our patients.

Dr. Patrick Agnew, DPM, DABPS  
Virginia Beach, VA

### Dear CMTA,

I have written this letter to inform other individuals who have CMT about my attempts to obtain private health insurance. I have been diagnosed with CMT since I was in grade school. I am now 37. I have always been covered under group health insurance plans with my employers. In fact, in the early 1980's, I was covered by Blue Shield in an individual policy. Last year, I decided to become self-employed again by starting my own environmental consulting company. I applied to Blue Cross for health insurance. After about a month, I got a letter stating that they would not provide coverage to me because, "Unfortunately, our medical underwriting guidelines prohibit us from offering enrollment to any applicant with a medical history of Charcot-Marie tooth disease." I used the exact spelling that was in the letter. Apparently, the individual reviewing my application either

made a typographical error, or thought I had a disease that affected my teeth.

I then called Blue Cross to find out more about their underwriting guidelines. According to Blue Cross, they have to follow their guidelines. They do not care whether you have any symptoms, or what the symptoms are, they only care if you have been diagnosed. I explained that many individuals who are diagnosed with CMT never have any symptoms, and the majority of those who do have symptoms live full lives with minimal health care. CMT does not impact one's life span, only one's lifestyle.

I then proposed to have my coverage specifically exclude costs associated with CMT. I personally am not concerned with the insignificant costs associated with CMT. I am concerned with the medical costs that could be associated with unrelated impacts such as car accidents, cancer and heart attack. Blue Cross would not even entertain that idea. I was then advised not to quit my job in order to maintain my health insurance because I would never get individual health insurance.

This did not seem fair or logical, and I was not going to let my life be dictated by one major health insurance company. I applied to other local insurance companies and was subsequently rejected by Blue Shield, Kaiser, and Health Plan of the Redwoods. Basically, individuals diagnosed with CMT have been black listed from obtaining health insurance from the above companies.

My persistence paid off and after about five months I did finally obtain standard health insurance coverage with the help of Mr. Sanford Bressick, of New York Life. He was able to obtain coverage for me from The Principal Mutual Life Insurance Company. The coverage is complete and includes CMT related costs.

Mr. Bressick is located at P.O. Box 1569, Santa Rosa, CA 95402, (707) 542-3462.

Very truly yours,  
D.J. Sebastopol, CA

*Editor's note: We invite responses from insurance companies who issue health insurance.*

(Continued on page 6)

## 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)  
Vincristine

**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 Hand Surgery, 3025 South Parker Road, Suite 3025, Aurora, CO 80014, 303/755-4588.

### Letters... continued

#### *Dear Friends,*

I hope I can call you friends even though we've never met before or even spoken over the phone. Since my husband and I have been attending support group meetings we've found CMT patients and families to be the friendliest kind of people you could meet.

I was interested in the letter of E.B.T. of Salem, OR, which was printed in your "Ask the Doctor" column. She inquired as to whether neurological problems such as mental retardation, dyslexia and attention deficit syndrome are related to CMT.

My first suspicion that one of my sons had CMT was when he had problems reading and reversed letters. My husband, who has CMT, had the same reading problems and so did a nephew with CMT. I thought this was too much of a coincidence, so, although my son had no physical symptoms, I took him to a local neurologist who was not sure he had CMT but told me that CMT and dyslexia seem to go together so often (in some families) that he felt there had to be some correlation although there had never been any proof of this.

That was about 20 years ago and my son now shows definite signs of CMT with foot drop very pronounced. He is very reluctant to inquire about it because it may cause problems with his health insurance, either now or later. It is interesting that females in the family who have CMT have had no reading problems. In fact, all read much more than the average American does nowadays.

R.G. Stratford, NJ

#### *Dear Editors,*

I was delighted to receive my first interesting and informative issue of the CMTA Report. I believe I received this because of my contribution through the Civilian Welfare Fund at work. I would like to continue to receive the report.

I am 53 years old and a CMT patient. My father, 85 years old, and my youngest son, 25 years old, are also CMT patients.

When I was in my early 20's, I was diagnosed as having had polio when I was very young. This diagnosis I've since determined was wrong. My father was never diagnosed as having CMT. When he was young, they said he had some disease whose name we can't re-

call. At the time, he had to learn to walk again. I had surgery to lengthen my Achilles' tendons, but I believe we were misdiagnosed because of the lack of knowledge of CMT at the time. My son was diagnosed while in high school, and since then, I have been, too.

I had to have a total knee replacement six years ago and during the pre-operative tests they found I had CMT.

I was very interested in your patient profile of Rhonda Serafini. The part about her losing her shoe was of much interest to me. I've never lost my shoe, but having the typical CMT foot, wide in front and narrow at the back, I have an extremely difficult time purchasing shoes...particularly something dressy.

I have to wear flat shoes for two reasons, weak ankles and bad balance caused by CMT and the artificial knee. For a while, I was fortunate to be able to purchase children's dress shoes in size 3 1/2 C with straps across the instep. The owner of the store went out of his way to find shoes for me, but he says he cannot order them in that size any longer. I guess there isn't much market for them so the manufacturers aren't making them. I could get shoes with ties, but who wants to wear tie shoes when you're dressed up. I may have CMT, but I still like to look nice.

I'd like to hear how others with CMT have solved this problem or what we can do to bring it to the attention of some shoe manufacturers.

Sincerely,  
R.K. Camp Douglas, WI

Editor's Note: It isn't often that we have an immediate answer to a patient's question, but in the same summer mail came the following information.

#### *Dear CMTA,*

I have noticed in your newsletter a lot of helpful information. I have always had difficulty finding shoes that fit. When I was diagnosed with CMT at age 16, I finally knew why. However, I still could only wear shoes that tied or buckled. Pumps would fall off my feet. When I could no longer fit into a child's shoe that had a strap, I avoided wearing dresses due to my lack of appropriate shoes. I believe God gave me sudden inspiration one day. I called a shoemaker from the phone book to ask if he could add ankle straps to pumps. The

(Continued on next page)

answer was yes! I excitedly bought my first pair of pumps and he put on matching straps for me at very little cost. It worked so well that I have had him add them to several pairs since then. The one problem I had was finding a nice pair of leather pumps that were wide enough to accommodate my spreading feet. I discovered that the brand "Rockport" meets this criteria. I hope this information is helpful to other readers who have difficulty buying shoes.

Sincerely,  
K.V. Walworth, NY

**Dear Editor,**

My son, now 5 1/2 years of age was diagnosed with CMT at age 14 months. He wears bilateral AFO's and walks quite well with them. His hand strength has decreased over the last 1 1/2 years. We have been trying to do everything possible for him but would like to hear from other parents of young CMT children. We would like to hear about all the accomplishments and struggles along the way, especially how children of this age learn to adapt.

Please write to us and send pictures if you can. We will return all letters.

Thank you.  
Kate Strong  
RP 4 Box 365  
Bloomington, IL 61704

**Dear CMTA,**

I just received the CMTA Report. Thank you so much!

In June 1991 my primary care doctor sent me to a neurologist when I went in for an exam of my legs. I had had leg cramps for years in my calves, but the leg spasms in my thighs were keeping me awake at night. I had never heard of CMT. When people ask why I sometimes use a cane, they think I am crazy when I say, "Charcot-Marie-Tooth."

My doctor has never treated it before. I've been exercising my feet and legs and also massaging them. I am 79 years old. I've always walked every day until I began staggering. I blamed it on getting older. I'm hoping the tape (Physical and Occupational Therapy) will assure me that I'm doing the proper exercise. I'm a widow and hope to stay in my home as long as I can. The MDA clinic told me to write my alphabet with my feet, so I do that and massage my feet and legs daily.

I can't believe so many doctors and nurses and health care professionals are so unaware of CMT.

Medicare and Blue Cross sent back the claim for the bloodwork I had done in the hospital at my doctor's request. They said they didn't pay for "dental" work! (Doctor Tooth, I guess!)

Thanks for listening.

J.H.

**Dear CMTA,**

I am a 27 year old woman with CMT and have only experienced limiting symptoms for about a year.

Since I have never seen alternative therapies mentioned in your newsletter, I am curious if any people with CMT have pursued treatments that are not considered mainstream: rolfing, yoga, special diet, acupuncture, etc.

List my address in your newsletter so that others may contact me if they have had any successes or failures that they would be willing to share with me.

Thanks for providing such an informative newsletter and for the work you do. You have immeasurably helped me combat the feelings of isolation and frustration they I sometimes experience from having CMT.

P.S. Based on my experience, I perceive a serious void concerning mental health options provided for the CMT patient. I believe there should be equal clinical emphasis on helping the patient emotionally adjust to the difficult changes and limitations brought about by having this disorder. The clinic I attend, for example, was unequipped to provide emotional support or even mental health referrals, a complaint I have heard echoed by a friend with multiple sclerosis. Perhaps more advocacy and research in this area might facilitate a more holistic and effective approach to the treatment of CMT.

Sincerely,  
Jeannie Garten  
213 Homestead Road  
Chapel Hill, NC 27516

*Editor's note: The CMTA continually sends information to physicians, hospitals, clinics, etc., If you would like to assist us in reaching facilities in your area, contact the CMTA and we will provide you with CMT brochures for distribution to your area institutions. We urge you to get involved.*

**Honoraria**

**In Honor Of**

Fred Smith  
Betty Johnson  
Christy Wagner  
Elia Atalla  
Fanny Ostrov

**By**

Margaret Walker  
Margaret Walker  
Pamela Gatfield  
Yacoub Atalla  
Tedi & Judi Friedberg  
M/M Haven Sharaf  
Anita Hoffman  
Stacey Hoffman  
James Goldsmith  
Hope Lariosa  
Cathyann Marcoux  
Joan Reivich  
Elizabeth Levy  
Rose Freed  
Robert Sand  
Pearl & Percy Sand  
Pauline Goldfarb  
Judy & Steve Fubin  
Diane & Michael Sand  
Minna Sand  
Sheila Marquis  
Joel & Nancy Weisberg  
Mark & Susanna Sand  
Janet Sand  
Stephen & Jean Sand  
Stephen & Rhonda Sand  
Dr. Maurice Sand  
Ida Sand  
Stephen Sand  
Judith Goldman  
Leslie & Richard Bosler  
Florence Survetnick  
Irwin & Sybil Whitman

Mrs. Hart Wurzburg  
The Corpuz Family  
M/M Thomas Phelan  
Rebecca Sand

**Memorials**

**In Memory Of**

Mrs. Eleene Agnew

**By**

Margaret Powell  
Karen Carlisle  
Carl & Eileen Jolly  
Vie & Sam Corl  
Blue Knights  
Motorcycle Club  
Mary & Patricia Haines  
Hirshella Young  
Hirshella Young  
Hirshella Young  
Hirshella Young  
M/M J.P. Hekkel  
Mary Brennan  
Ruth Feen  
Drucilla Lake  
Mrs. O'Day Baker  
Yacoub Atalla  
M/M Wm Wheat, Sr.  
Marsha Wheat  
Jackie Carlson  
Madelynn Brown  
Karol Hitt  
CMTA Bd. of Directors  
Janet Phy  
Joan Vater  
Joan Vater  
Joan Vater  
Irene & Len McMahon

Bernie Young  
Rose Winnick  
Murray Winnick  
Isadore Winnick  
Marcia Hekkel  
John W. Brennan  
Hascal Feen  
Leo Breeden  
O'Day Baker  
Shafiqua Atalla  
William Wheat, Jr.

Edwin Carlson  
Freddie Rothstein  
Dymphna Lovelace

George Phy  
Gerge Nilson  
Dr. Peter Ciano  
Daniel Guertin  
Angelo Theoharis

**Alert, Alert**

We have been reviewing our records, and by now everyone in our database should have received a letter asking for a membership renewal or a confirmation of the desire to continue receiving our newsletter. If we have not heard from you, your name will be removed from our mailing list as of January 1, 1993.

# A SUMMARY OF CURRENT CMT GENE RESEARCH



## THE HAND in Charcot-Marie-Tooth Disease: A Review

by Donald J. Perrella

In an event unprecedented in CMT research, back-to-back articles in the scientific journal *Nature Genetics* by four separate groups of researchers reported the identification and location of what all believe to be the candidate gene responsible for CMT1A, the most common subtype of CMT. In each case the scientists reported direct evidence locating the human peripheral myelin protein (PMP-22) gene to a previously discovered duplicated region of DNA on chromosome 17. The first article reported the isolation of cDNA and genomic clones for human peripheral myelin protein (PMP-22). This gene has been mapped to the 17p11.2-17p12 region of chromosome 17. It is expressed in high levels in peripheral nervous tissue and is duplicated, but not disrupted, in CMT1A patients. This increased gene dosage is believed to be at least partially responsible for the demyelinating neuropathy seen in CMT1A.

The authors of the second article reported that the PMP-22 gene is located within the CMT1A DNA duplication, which is a direct repeat and does not interrupt the coding region of PMP-22. Expression of PMP-22 in CMT1A fibroblasts is similar to expression in control fibroblasts. Increased gene dosage or altered PMP-22 expression in the peripheral nervous system are therefore possible mechanisms by which the PMP-22 gene is involved in CMT1A.

In the third article the researchers reported that the human PMP-22 gene is

contained within the CMT1A duplication and this increased dosage of the PMP-22 gene may be the cause of the CMT1A neuropathy.

The scientists authoring the fourth article reported the construction of a partial yeast artificial gene chromosome contig spanning the CMT1A gene region and the mapping of the PMP-22 gene to the duplicated region. The scientists stated that these observations further implicate PMP-22 as a candidate gene for CMT1A, and suggest that over-expression of this gene may be one mechanism that produces the CMT1A phenotype.

These four articles are the culmination of an understanding of the molecular pathology of CMT that began in 1989 when Vance *et al* linked CMT1A to markers on the proximal short arm of chromosome 17.

Then in 1991 Lupski *et al* and Raeymaekers *et al* found that certain markers were duplicated in CMT1A patients and that the duplication was linked to the CMT1A phenotype. Lupski *et al* also produced data suggesting that the duplication was of significant size. The *Trembler (Tr)* mouse is an animal model for CMT. *Tr* mice have hypomyelination in the peripheral nervous system and continuing Schwann cell proliferation. In two 1992 papers, Suter *et al* described a point mutation (a change in only a single nucleic acid) in *Tr* mice in the gene for peripheral myelin protein-22 (PMP-22) and a second allelic mutation in PMP-22 in *Trembler gmice*. This was sufficient reason to prompt the research carried out by the four groups whose results were published together in *Nature Genetics* strongly suggesting that overexpression of the PMP-22 gene is, at least partly, the cause of CMT1A.

Formal proof of this hypothesis involves excluding a possible effect of other genes in the region of the duplicated gene or by showing that the few CMT1A patients who appear to lack the gene duplication may, like the *Tr* mice, contain a defect within the PMP-22 gene itself. §

*Editor's Note: If any reader would like the bibliography used to assemble this article, please write to the CMTA office requesting it.*

In the May 2, 1991 issue of, *The Journal of Hand Surgery*, Drs. M. J. Miller, L.L. Williams, S.L. Slack and J.F. Nappi reported on a study they did on the CMT hand. The authors utilized both patient questionnaires and physical examinations. The results of the survey questions were: 1. hand symptoms began at an average of 19 years after onset of the disease 2. a broad range of motor and sensory changes were identified (75% motor deficits, 50% loss of strength, 15.6% dexterity, trembling 4.7%, cramping 4.7%). In 23% of the patients sensory changes were the most disturbing symptoms. Upon testing 98% of the patients were found to have motor abnormalities.

Regarding progression of the disease the authors suggest that most of the hand deficit occurs in the first five to ten years of hand involvement and then remains relatively stable. In their discussion of the progression of CMT the authors noted, "Carpal tunnel syndrome and other compressive neuropathies can mimic CMT and patients with this disorder may also have arthritis and tendinitis causing hand complaints. Such everyday disorders must be excluded when managing patients with neuropathy."

In discussing therapies the authors refer to surgical options to improve hand function (a little known option for the CMT patient), hand exercises, and the use of adaptive devices (aids for daily living). The authors commented upon how few patients were aware of treatment and rehabilitative procedures including surgical treatments. (*Editor's note: See CMT FACTS, VOL. I, pg. 12*) To quote the authors, "This is obviously a problem of patient education. It is necessary to make our colleagues who are not regularly involved in hand rehabilitation more aware of the ways of helping such patients. *Editor's note: For a reprint of this article write to Michael J. Miller, MD, Box 62, UTMDACC, 1515 Holcombe Blvd, Houston, TX 77030, USA. §*

### Coming Soon - Survey Results

Dr. Rhonda Jones will publish in the Winter newsletter preliminary findings from the survey she did in the Spring 1992 issue of *The CMTA Report*.

**The CMTA Report** is published by the Charcot-Marie-Tooth Association, a registered non-profit 501 (C)(3) health organization. The newsletter is co-edited by Carol Hitt and Pat Dreibein. The layout is by Chesapeake Bay Design, 1418 Ticonderoga Ave., Ridgecrest, CA 93555.

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.

# Support Group Notes

*A primary goal of the CMTA is to become a truly successful advocate for those with CMT. Its message must reach the patients, their families, and the medical and research communities. Patient family support groups help carry out this function.*

*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 notify the CMTA by mail or call 215-499-7486.*

*Perhaps there is a group meeting near you. You are cordially invited to join these groups in their upcoming events.*



Alabama - Greater Tennessee Valley  
Bill Porter 205-386-6579W; 205-767-4181  
Meets at ECM Hospital, Florence, AL

California - Los Angeles Area  
Oxnard-Thousand Oaks  
Janice Hagadorn (805) 985-7332

Adelanta (High Desert)  
MaryL Michels (619) 246-7807

Canyon Country - Saugus  
Sheila Levitch (805) 254-5322  
Denise Miller (805) 251-44537

California - San Diego  
Gary Oleze (619) 944-0550

California - San Francisco  
David Berger (415) 491-4801

California - Santa Rosa  
Freda K. Brown (707) 573-0181

Colorado - Denver Area  
Dr. Gregory Stilwell (719) 594-9920

Florida - Orlando Area  
Carole Wray (407) 788-7427  
Meets 3rd Sat. every other month  
Metro West Church of the Nazarene  
Orlando, FL

Florida - South  
Robyn Cohen (407) 622-5829

Massachusetts - Boston  
Eunice Cohen (617) 894-9510

Michigan - Brooklyn  
Robert D. Allard (517) 592-5351

Michigan - Detroit  
Suzanne Tarpinian (313) 883-1123

Missouri - Kansas City  
Sandra Toland (816) 756-2020

New Jersey - Central  
Janet Saleh (908) 281-6289  
Somerset Medical Center  
Sommerville, NJ 08876

New Jersey - Northern  
Teresa Daino (201) 934-6241  
Meetings: Englewood Hospital  
Clinic Conference Room  
350 Engle Street, Englewood, NJ

New Jersey - Millville Area  
Linda Muhlig (609) 327-4392

New York - Long Island  
Lauren Ugel (516) 433-5116

New York - Rochester  
Neale Bachmann (716) 554-6644  
Bernice Roll (716) 584-3585

New York - Westchester County  
Kay Flynn (914) 793-4710

North Carolina - Eastern  
Susan Salzberg (919) 967-3118  
(919) 286-0411 (x6586) days  
Durham VA Medical Center

Ohio - Cleveland  
Norma Markowitz (216) 247-8785

Pennsylvania - Delaware Valley  
Rex Morgan, Jr. (215) 672-4169

Texas - Greater Dallas Area  
Dr. Karen Edelson, D.P.M. 214/542-0048

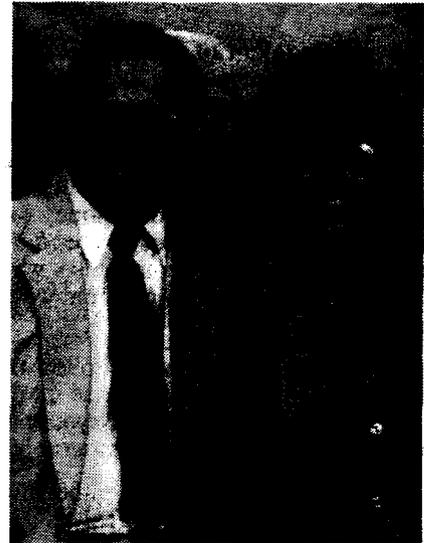
Utah - Salt Lake City  
Marlene Russell (801) 966-7563 home  
(801) 565-1212 work

Virginia - Tidewater Area  
Mary Jane King (804) 591-0516  
Thelma Terry (804) 838-3279

Virginia - Richmond Area  
Dennis Breckenmaker (814) 748-9021  
Steve Firestone (804) 745-4123

West Virginia - Central  
Joan Plant (304) 636-7153 (after 6pm)

## support group corner



The Porters

Bill Porter is the leader of the new support group which he formed in the Greater Tennessee Valley. Bill lives in Florence, Alabama, and works as a metallographer for a large aluminum producer. He spends much of his time at work analyzing defects in beverage cans and aluminum sheet using optical and scanning electron microscopes. Bill and his wife, Bonnie, have three daughters (12, 13 and 23 years old) who have not yet been tested for CMT. Two of his daughters are sports-oriented and are active in competitive swimming and track. His wife is a kindergarten teacher and a master jeweler for Premier Designs. The Porters are sports directors for their school and are actively involved in their church. CMT has been part of Bill's life since he and his sister and brother were diagnosed 8 years ago. Despite ankle-foot braces, arthritis and back problems, Bill still manages to do most of the things he enjoys. Bill decided to start the support group mainly because he did not know anyone with CMT except his own relatives in Virginia. No one in his area seemed to be very familiar with the disorder. The first meeting was a great success, with 22 people attending. Some drove 6 hours from Mississippi to be at the meeting. Nearly everyone participated in the discussion following the presentation by neurologist, sDr. Dina Jannun. Bill is urging the CMTA members from Tennessee to give him a call and to try to attend the next meeting. No one from Tennessee came to the first support group session and he would like to meet some of you. §

The inaugural meeting of the new CMTA Support Group of South Florida was September 26, 1992 in Saunderson's Hall, St. Mary's Hospital, West Palm Beach, Florida. The group was treated to lunch by Frank Crohn and the Cresthaven Chiropractic Center and was privileged to hear Dr. Walter Bradley, D.M., F.R.C.P., Professor and Chairman of the Department of Neurology, University of Miami, speak on the topic of "Neurology and CMT." Thirty-five people attended this meeting.

If you live in South Florida, and missed this first meeting, please call Robyn Cohen and get on the mailing list for the next support group meeting (1-407-622-5829). §

## TALENT ABOUNDS!

The artistic talent of the members of the CMTA is astonishing and one that we in the organization would like to promote. In the past, we have asked for artists to send us sketches that would be suitable for use in creating notecards that could be sold to raise money for our research fund. We continue to be interested in hearing from artists who have an interest in having their work featured on notecards.

This summer we received the following letter from a sculptor who has CMT and whose accomplishments you can see in the accompanying photograph.

"Please find enclosed my membership and contribution. I just received my first copy of your newsletter from my parents, and I found it very encouraging and informative.

I am 25 and have been diagnosed with CMT since I was 14. I went to many doctors, one who even told my mother and I that I had a terminal muscle disease and would die in pain, bedridden, before age 25. He later admitted to accidentally having read about the disorder listed next to CMT in his medical book!

Well, everything has gotten much better since that point. I started my career in sculpture five years ago, and moved from my home state of Nevada to the sculpture capital of the United States, Loveland, Colorado, two years ago.

Since my move and employment at an art foundry, my career has taken off. I have just finished my first monument, a lifesize whitetail deer, and I will be buying my first home soon. I never thought I could get this far, but when I concentrate on what I can do, and not on what I can't, I can achieve anything I want.

I am affected by CMT in my legs and hands and arms. I use a crutch all the time, and my hands are noticeably thin and weak.

In your letters to the Editor, someone mentioned that he doesn't receive any attention from the opposite sex because of his CMT. He is around the wrong people! My experience has been completely positive, with my CMT weeding out all the jerks and shallow people. The people who choose to be with me are intelligent, sensitive beings who never see me as less than an equal. Right now, I have the most wonderful and handsome boyfriend I've ever had.



I have asked many of my able-bodied companions why they choose to be with me over a "healthy" person, and they usually look amazed and reply that they like me for what's inside, and they don't even notice my crutch and funny walk anymore.

If you think about it the right way, a disability can be a blessing and a teacher. I know that if I weren't affected, I would not be who I am now. Instead of sculpting and appreciating the beauty in simple things and pleasures, I would be hanging out in the mall or cruising around trying to look important. CMT makes me appreciate life on its simplest terms, and to savor everything.

Sarah Rose  
Loveland, CO

*We were so impressed with Sarah's letter and her work that we have decided to propose the creation of THE ARTIST'S PROJECT whereby artists could donate a piece of their work to be auctioned by the CMTA with the proceeds going to the research fund.*

*If you are a professional artist or an accomplished amateur, we would love to hear from you. Please send us a brief biography, a photograph (if possible) and a picture of your work. §*

### LETTERS

**We want to hear from YOU!**

Write us at:  
Letters / The CMTA  
601 Upland Avenue  
Upland, PA 19015



## Ask the Doctor

### Dear Doctor:

*I am a mother of three year old boy. I delivered by Caesarean section, and at the request of my neurologist, I had general anesthesia. I am now pregnant with my second child and would like to be given the choice of epidural anesthesia. Can you tell me if there is any evidence to suggest that epidural or spinal anesthesia may risk progression of the symptoms of CMT?*

S.S., CA

### The Doctor responds:

"To my knowledge, there have been no studies of the risk of spinal anesthesia in patients with Charcot-Marie-Tooth disease. However, there are some important theoretical considerations which need to be taken into account before proceeding with spinal anesthesia. Firstly, the nerve roots in patients with CMT are usually enlarged in the same way as other peripheral nerves are. This has two potential consequences for spinal anesthesia. First of all, there is a greatly increased likelihood that a needle inserted into the spine will hit one of the nerve roots which is almost certain to produce pain and carries a very small but identifiable, risk of nerve injury. Secondly, because of the presence of multiple enlarged nerve roots in the spinal space, flow of the anesthetic agent may be impeded and inadequate anesthesia achieved. I believe that these theoretical risks to spinal anesthesia in patients with CMT are very small and need to be balanced against the patients' wishes as well as against the generally greater risks of general anesthesia. I would emphasize, that even if one of these above mentioned problems does arise, there is nothing to suggest, either from prior studies or on theoretical grounds, that this would contribute in any way to the acceleration of the progression of symptoms in CMT."

### Dear Doctor:

*A patient from Kansas writes: "I am having a problem that I wonder if other CMT patients have. I am experiencing sores that are not open, but that are from the inside out. These sores burn and itch and I have great difficulty wearing shoes. If I put any type of pressure on them, no matter how little, I just can't stand the burning, searing pain. What can be done about this? Is this common?"*

(continued on next page)

**A podiatrist from the Medical Advisory Board replies:**

"People with Charcot-Marie-Tooth disease frequently have structural foot problems because of muscles imbalances. Most commonly, this is reflected in a high arch foot type. The distribution of pressures across the ball of the foot in these patients is quite exaggerated. Frequently patients will develop points of significant pressure which may result in ulceration, but which more commonly create areas of very thickened skin and significant pain. Most commonly, these pressure areas are across the ball of the foot because of the shape of the foot, and also because of the foot drop condition which may accompany CMT and which results in greater pressures on the ball of the foot. The pressure areas may be balanced out with a full length type of insole which can help to distribute pressures more evenly across the whole foot. Depending upon the severity of the structural deformity and the degree of muscle imbalance, other kinds of procedures such as surgery may be helpful. Most patients can be helped to a very significant degree by one form of therapy or another.

*A patient asks about the drugs on the neurotoxic drug list: "What do these drugs (on the neurotoxic drug list) do that is harmful to the CMT patient? Do all of the listed drugs have the same harmful effects or do they differ? Do the harmful effects go away once use of the drug is terminated or are the effects more long lasting?"*

**A Medical Advisory Board neurologist replies:**

A complete answer to this question would require an analysis of each one of the drugs reported to cause neuropathy to see what experimental data exists to document the extent of the toxicity and the part of the nerve that is actually damaged.

All the clinicians and patients need to know at this point is that drugs are potentially toxic to peripheral nerves and that damage is potentially permanent. If a drug happens to be toxic to axons i.e. the nerve fibers, then improvement must await regrowth of the fiber, a process that can take many months or years and wind up being inefficient and incomplete.

In contrast, if a toxic substance damages the myelin sheath, then repair might be completed in several months and full function returned. Certain

## 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
<input type="checkbox"/> Other	

**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)
- Booklet - CMT FACTS (\$3)
- 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) (free to members with self addressed stamped business envelope) please list states: \_\_\_\_\_
- Medical Brochure - CMT (gray brochure) (free to members with self addressed stamped business envelope)

**Total amount enclosed:** \_\_\_\_\_ Contributions are tax deductible.  
Please make checks payable to the CMTA.

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.

drugs affect the motor fibers preferentially and do not cause sensory symptoms. An example of this is Dapsone. Others, such as Cis-platinum, appear to preferentially affect the sensory system and cause damage to the dorsal root ganglion cell which controls reception of all aspects of sensation from the periphery. In my own practice, I've only seen one from the list actually cause damage in Charcot-Marie-Tooth and that drug is Macrodantin, a commonly used urinary antibiotic, where several members of one family devel-

oped sensory symptoms after several weeks on this drug. Only after they reported the symptoms were they actually examined and diagnosed with CMT. Finally, patients and physicians interested in details on any one of the drugs should look it up and should not consider the medical literature to be a private affair. It may be a little difficult at first, but editors of modern medical articles are increasingly interested in simple English sentences and logically stated arguments. §

**CMTA Recognizes  
The 1992 Rebecca Sand  
Volunteer of the Year**



**Mary Beeler**

The CMTA is pleased to announce that the Volunteer of the Year award is being shared this year by two long-time supporters of this organization. The first recipient of the Volunteer of the Year award is Mary Beeler, former support group leader in Orlando, Florida. Mary began the Orlando group in 1988 after attending a regional conference and being "talked into" getting a support group started. No one had a more loyal following or was more highly regarded as a leader than Mary. She has just recently stepped down as the leader because of the demands of her job and because she has returned to school. Mary was exemplary in the consistency with which she held her meetings (one every other month since the beginning) and the spirit of infectious good humor which she imparted to her group members. The CMTA is honored to have had Mary as its representative and now as its recipient of the Volunteer of the Year award.

The second recipient is Walter Kelly, Jr. of Boston, Massachusetts. Walt has done the bulk of his volunteerism for this office by editing and producing the new tapes for our CMT tape purchase program. Through Walt's efforts, we have been able to pair presentations from two different conferences on one tape, such as the Physical Therapy and Occupational Therapy presentations. Walt has not only edited and produced the originals of the current tapes, but he has copied and produced more than one hundred tapes that have been purchased by our members. Working from his home, Walt has performed a service which all of our membership benefits from. The CMTA is honored to present the second 1992 Volunteer of the Year award to Walter Kelly, Jr.

Volunteers like Mary and Walt are the backbone of the CMTA, and they are the reason we continue to grow and are able to serve the CMT community. §

## CMT...

- ..... is the most common inherited neurological disease, 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, occasional partial sight and/or hearing loss problems 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, affecting half the children in a family with one CMT parent.
- ..... may become worse if certain neurotoxic drugs are taken.
- ..... can vary greatly in severity, even within the same family.
- ..... is the focus of significant genetic research, bringing us closer to answering the CMT enigma.

## ***THE CMTA REPORT***

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

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

Crozer Mills Enterprise Center  
601 Upland Avenue  
Upland, PA 19015

**TO:**

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