

The CMTA Report

JUNE 2001

Vol. 16, No. 3 ISBN #1067-0181

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

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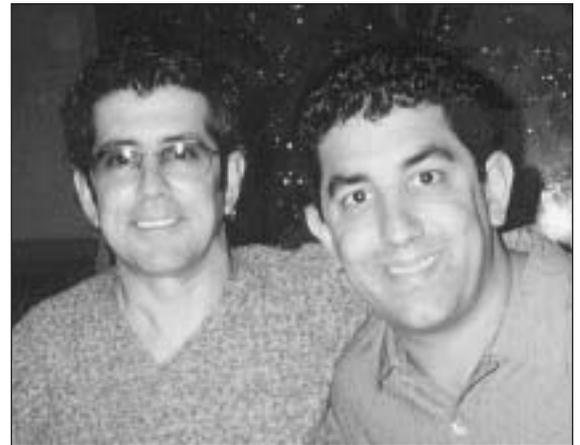
Brothers Honor Father's Memory with Golf Tournament

On the twenty-fifth anniversary of their father's death, Christopher and John Scarduzio of suburban Philadelphia are planning a golf tournament in his honor to benefit the Charcot-Marie-Tooth Association. Both the father, John J. Scarduzio, and his son, John A. Scarduzio, were diagnosed with the disorder. Other family members are also affected. To honor his father's memory and to support research that might benefit his other affected family members, Christopher Scarduzio originated the fundraising idea.

The golf outing will be held on Monday, September 10, 2001, at the Springfield Country Club, 400 West Sproul Road, Springfield, PA 19064. A foursome for the event costs \$1,000 and a single entry costs \$250. Foursomes can be arranged for those who do not have a registered foursome. Patrons to sponsor a hole at \$500 are actively being sought. Sponsors will have their name (or their company name) prominently displayed on the tee box.

The entry fee will entitle a participant to 18 holes of championship golf, a riding cart, a bag drop attendant, use of the clubhouse lounges, showers and locker rooms, boxed homemade lunch, refreshments throughout the rounds, grand buffet, open bar and an awards reception following play.

The day will begin with registration at 9:00 AM and a start time of 10:00 AM. The day will conclude with a 7:00 PM reception and prize announcements at 7:30 PM. There will be valuable prizes for longest drive, closest to the pin, low gross and best low ball of a foursome. The most coveted prize is a 2001 BMW for a hole-in-one, which is being sponsored by Princeton BMW of New Jersey.



John and Christopher Scarduzio are organizing a golf tournament in September to raise money for CMT research.

Christopher Scarduzio, the organizer of this event, is the Executive Chef at Brasserie Perrier in Philadelphia and has also been a chef at Le Bec Fin, a renowned Philadelphia culinary landmark. He will be donating the boxed lunches from his restaurant. Some of the prizes for the chance drawings will include a meal prepared by Chris in the winner's home, meals at the Brasserie and vintage wines.

Because this event is being held close to our headquarters, the CMTA encourages our local members to support this fundraising event in any way they can. People can attend the dinner only for \$100, sponsor a hole, donate a raffle item or simply make a donation to support the effort. Please call the office (1-800-606-2682) if you have questions about the outing or the way in which you might participate.

The CMTA: Leading the Way in CMT Research

The CMTA.... Living our Mission

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

Vision Statement:
Dedicated to generating hope.

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

Last month, as part of our educational outreach program, we staffed a booth at the American Academy of Neurology Annual Meeting in Philadelphia. While there we also met with the CMTA Medical Advisory Board. As anticipated, the CMT North American Database generated much excitement and discussion. In later conversations with members of the Medical Advisory Board, several expressed opinions that they believe CMT has more serious complications than has been previously thought. Because of our involvement with many CMT patients and their families, we at the CMTA have felt this way for a long time, but, until now, we have not had the resources to collect enough solid evidence to support these beliefs.

The CMT North American Database is the vehicle that will provide us with the information that is needed to support, or maybe even discount, these beliefs. It may even produce some surprises. However, because this project will

involve the largest number of CMT patients that has ever participated in a CMT research study, it will provide clinicians and researchers with a more complete picture of the severity and progression of CMT. It is indeed the most important research project that has ever taken place in the history of CMT.

As was related in the last issue of *The CMTA Report*, not only will the information gleaned in this study allow us to finally say with certainty "This is what CMT actually is," it will also provide us with a map of the various directions CMT research should take in the coming years; speed up the pace of CMT research; stimulate research interest; and attract more research funding—all of which are necessary and long overdue.

To ensure the quality of the research, an Institutional Review Board comprising CMT Medical Advisory Board Members has been formed. The task of this board is to evaluate research proposals and decide who will have access to the information culled through the Database.

We stress again that there are a number of safeguards in place to protect you. From the beginning, patient confidentiality has been and will continue to be a major concern. All participants are assigned a number and no names will be given out to any researcher without permission.

This project is the most important research project that the CMTA has ever undertaken. We not only need you to participate by enrolling in the study and filling out the forms, but, in order to keep this project going for the next two years, we need your financial support as well.

The Buuck Family Foundation has generously given us a \$60,000 grant to establish and maintain the CMT North American Database for the first year. The CMT Clinic at Wayne State University also received a \$31,000 grant to pay for a research assistant to work with the project for 2001. However, these grants were given with a challenge—that the CMTA raise enough money to keep the Database funded for the second and third years. We estimate that the second year will cost at least \$91,000; the third year at least \$101,000. Contributions to the CMTA Research Fund over the next few years will be of the utmost importance.



Chairman Ann Beyer and Vice-President Vincent Bertolino attended the American Academy of Neurology Meeting in Philadelphia in May.



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

2001 Research Fund Progress



Contributions to the CMT Research Fund will allow the North American Database project to continue for the next two years and fund postdoctoral grants.

Besides funding the CMT Database, we would also like to continue with our postdoctoral research grants. The CMTA is leading the way in CMT research. Researchers from top institutions are applying for and receiving our grants. Our CMTA Fellows are carrying out cutting-edge research. This year we received seven outstanding proposals for our CMTA Fellowships. We were able to fund only five of them. However, this is the highest number of postdoctoral grants we have ever funded. And their findings will be presented at the American Neurological Association satellite forum this fall. The CMTA is one of the very few institutions that funds CMT research.

A number of you have asked me why so few institutions fund CMT research, and why we aren't going out to drug companies to get research funds. The answer is that the funds are not out there. Having a disease that has been designated as a rare or orphan disease hurts us deeply. In fact, it is a kiss of death. A rare or orphan disease is a designation instituted by pharmaceutical manufacturers for any disease that affects under 200,000 people in the United States. What this means is that the condition affects too few people to make it profitable to engage in research to develop treatments. I use the phrase "kiss of death" because if pharmaceutical manufacturers decide that a disease is not worthy of research attention, it will not generate research or medical interest either.

(continued on page 4)

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

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

VISA MasterCard American Express

Card Number _____ Expiration Date _____

Signature _____

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

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



VISIT OUR HOMEPAGE

at

www.charcot-marie-tooth.org

US Boost for CMT Research in Australia

(Editor's note: The following article appeared in the Autumn 2001, Volume 38, CMT Australia Newsletter. It discusses the grant made by the CMTA to an Australian research team.)

Research into the day-to-day effects of living with CMT has received a boost, with the appointment in March of a full-time CMT research fellow at the Children's Hospital at Westmead (formerly known as the New Children's Hospital).

The Moore Postdoctoral Fellowship, funded by the CMTA in the US, provides funds for a program of research aimed at evaluating some of the effects of the disease on sufferer's leg and foot function, and the impact on daily life that results from the functional changes. The award was made to a team including Anthony Redmond, the new research fellow, well-known Professors Robert Ouvier and Garth Nicholson and physiotherapy lecturer Dr. Kathryn Refshauge of the University of Sydney. "We are particularly pleased," said Anthony, "that funding has been obtained for a study which is of practical use here and now."

"Currently a great deal of emphasis is placed on the genetic and molecular aspects of research into neuromuscular disorders. Of course, these efforts are important, as in time, they should lead to the discovery of a real cure for diseases such as CMT. In the meantime, however, we are very happy that programs of work aimed at improving the lives of people with the disorder now are also being supported."

The aims of the research program are: 1) to identify or develop measures which can be used to accurately define the physical and functional changes occurring in CMT and similar neuromuscular diseases, 2) to describe in detail, the natural progress of functional aspects of CMT, and 3) to plan and evaluate targeted treatments aimed at addressing the specific pattern of nerve and muscle changes seen in CMT.

The main study this year is an extension of the trial already being conducted by Dr. Refshauge and Associate Professor Garth Nicholson at Concord Hospital. Involvement in this trial involves wearing a splint on one leg at night for a period of 3 months. During and after the treatment period, the study team will evaluate in detail the effects of the treatment. The detailed

evaluations will give the researchers important information about the effects of CMT as well as a better idea of how to plan effective treatment approaches. As part of the study, the effect of age on the effectiveness of the treatment will be considered so they are asking the help of child volunteers (age 5-15) and older children and adults up to age 30.

"The lack of studies like these is precisely the reason why the success of current treatments is so patchy. Good quality research is the only way to ensure that the next generation of children with CMT receive better care than we have been able to provide in the past."

LEADING THE WAY

(Continued from page 3)

Maybe through information gained in the Database, we will find out that CMT is not rare at all. That maybe it does affect more than 200,000 people. Think what that would mean for all of us!

In order to continue these research programs, we need your support. Our Annual Research Appeal letter went out several weeks ago. So far you have given almost \$32,000. We would like to raise over \$200,000 to keep both the Database and CMTA Fellows projects functioning next year. In order to do so, we need your financial support. I believe we can raise the funds.

The purpose of this Database and the post-doctoral fellow programs is to help all of us who are affected by CMT. Almost everyone on the CMTA Board of Directors is affected by CMT, and we give many hours each week to make a difference. We also contribute to the CMTA because we believe in what we're doing. The CMTA is leading the way in CMT Research. But the CMTA is not just an office in Pennsylvania and a Board of Directors and a Medical Advisory Board: it is all of us working together to make a difference.

We are deeply committed to finding treatments, and eventually a cure, for CMT. But we cannot do this alone. We need your financial support.

Working together we can cure CMT!

Message From the President...

During my first six months as President of the Charcot-Marie-Tooth Association, a number of significant goals have been accomplished within our organization. This year we've funded more research grants than ever before and we've actively supported the formation of the North American CMT Database. Of great importance was the long-awaited first Support Group Leaders' meeting in St. Louis in April. This weekend meeting brought together a group of enthusiastic leaders or their representatives to discuss the goals and objectives of the CMTA and the ways in which support groups and the organization can help each other.

One important subject discussed during the meeting was the need to find a well-known National Spokesperson. It is absolutely impera-

tive that the name Charcot-Marie-Tooth (CMT) become recognized as a common peripheral neuropathy and not "Charcot Foot", and certainly not a dental disease. A recent Sunday newspaper article about multiple sclerosis, highlighted TV talk show host, Montel Williams, who has MS. Whether it's Montel Williams talking about MS, or Michael J. Fox about Parkinsons, these well known and highly visible personalities have brought knowledge of their disorders to the general population. I would like to challenge our CMTA members and their families, as well as others interested in our disorder, to join in a search for a well known spokesperson so that the next time one of us tells someone that we have CMT, the answer won't be, "What's that?"

—Ardith Fetterolf

Students Increase Knowledge of CMT



Four physical therapy students from Widener University, Chester, PA, made the decision to do research and write a paper on a neuromuscular disease. They narrowed their choices to the problems of the peripheral nervous system and chose Charcot-Marie-Tooth disorders because they believed that what they learned would ultimately help them in treating patients. Their paper, written for a histology class, was entitled "The Role of Gap Junctions and Connexin 32 Protein in Charcot-Marie-Tooth Disease." Pictured above, in the CMTA's office, are, left to right, Leigh Walthall, Brenda Ashby, Mona Khan, and Renee Zavros.

The Psychiatric Aspects of Peripheral Neuropathy

By **SCOTT BERMAN, MD**

(as presented at the Peripheral Neuropathy Symposium in Allentown, PA)

(Editor's note: Dr. Berman suffers from CIDP, chronic inflammatory demyelinating polyneuropathy, and spoke based on both his own personal experiences as well as his clinical practice.)

There are many effects of chronic illness (such as all of the neuropathies) which are not specifically physical in nature. In fact, many psychological problems arise as secondary issues in chronic illness. The effects of chronic illness are depression, anxiety, anger, denial, fatigue, sleep disturbance, sexual dysfunction, family stress, isolation and loss of social function.

Here we will consider three of those effects: depression, anxiety and fatigue.

Depression is an independent cause of death and disability in chronic illness. It not only causes mental problems, but it also contributes to a poorer prognosis physically, if left untreated. Depression is one of the top 5 disabling and costly of all diseases. In 20 years, it will be second only to heart disease in its cost to society. However, in spite of the fact that it is often left untreated, depression is highly treatable, even in the presence of multiple severe chronic illnesses.

If depression is so treatable, why is it undertreated? In many cases, the patient is unaware of his depression or he is embarrassed to seek help. Physicians tend to underdiagnose the condition, often because they have the misconception that depression is a normal part of chronic

illness and not a separate illness all its own. In some cases, both patients and doctors are fearful of a referral to a “shrink!”

How can a person tell if he or she is depressed? The symptoms of depression are easy to spot. They are either an increased or decreased ability to sleep, a lack of interest and loss of pleasure in activities, a feeling of guilt or worthlessness, a decrease in energy, a decreased ability to concentrate or remember, increased or decreased appetite, psychomotor retardation or agitation and, most frighteningly, suicidal thoughts. About 10% of all depressed people develop suicidal tendencies. If someone has five or more of these symptoms, lasting 2 or more weeks, he would be considered clinically depressed.

There are treatments for depression and one or more of them should work for every person. The first treatment that is usually tried is antidepressants. They will work, if they are used in a high enough dose and for a long enough time. Unfortunately, they are often given in a dose which is too low and for too brief a period of time. Psychotherapy, which is either cognitive-behavioral or interpersonal, is virtually unheard of in this time of managed care because it is simply too expensive. Relaxation techniques, such as deep breathing, can be effective in helping to release tense muscles and lead to an overall better feeling. Depression is often fueled by grief and disputes in the family, so group, cou-

CLINICAL TRIAL FOR CMT TYPE 1 STARTS AT OHIO STATE

Dr. Zarife Sahenk, Professor of Neurology, Ohio State University, is studying the “Effects of BDNF and NT-3 treatment on CMT1A xenographs” with a grant from The Neuropathy Association. Her studies show that NT-3 improves axon regeneration and remyelination in CMT1A nerve segments. She is doing a pilot clinical trial using diagnosed type 1 patients in the Ohio area. Her theory is that the Schwann cells in peripheral nerves elaborate the myelin sheath and support the normal functioning of the axons. When the Schwann cells are defective as in Charcot-Marie-Tooth Disease, the axons degenerate. Dr. Sahenk and her colleagues propose to show that certain neurotrophic factors, such as NT3 can make up for the abnormal Schwann cells, and provide support to the axons. If true, these neurotrophic factors could preserve axonal function in CMT neuropathies.

ples and family therapy can also be helpful.

The second major psychological result of a chronic illness is anxiety. Anxiety is a generalized sense of tension and worry. It arises from a fear of relapse, a fear of disability, a fear of death, a fear of isolation and a fear of stigmatization resulting from the visible signs of illness, such as braces, canes and deformed limbs.

There are many treatments for anxiety, but most people choose nothing. Common treatments include medications, relaxation training, psychotherapy, faith and support groups. We will discuss the most commonly used medications. Many people fear the habit-forming nature of anti-anxiety drugs such as Valium, Ativan, Xanax or Klonopin, but the difference between dose and overdose is extremely high. Sleeping pills such as Sonata, which provides four hours of good sleep, and is, therefore, useful for a 2 AM dose, and Ambien, Restoril, or Benadryl can provide the restorative sleep that is so essential for people with an ongoing disease. The common antidepressants with anti-anxiety effects are Remeron, Serzone, Paxil, and Effexor. Neurontin helps with both anxiety and pain and Risperdal and Zyprexa cause fewer neuromuscular side effects than some of the others.

Fatigue is the final condition related to chronic illness which we will consider. A report in *Neurology*, November 1999, reported that severe fatigue was reported in 80% of neuropathy patients. The fatigue was independent of the patient's motor or sensory symptoms. Fatigue was rated as one of the three most disabling symptoms of the person's disease. Depending on what type of neuropathy one has, treatment might include any therapy or device which would modify the condition. (For instance, using braces to help in walking could reduce fatigue.) Exercise and physical therapy often serve to regenerate a patient and to strengthen muscles, which can make tasks less draining. Finally, adequate rest periods are a must and should be planned into each day. Rest, combined with adequate hydration and nutrition, can help control fatigue.

Conclusions: Neuropathy often leads to depression, anxiety and fatigue. Psychiatric care of these conditions can improve the overall quality of one's life. For psychiatric care to be effective, the patient and referring doctor must not leave the patient with the implication that the disease or the pain is "not real." The psychiatrist must be part of the treatment team and carefully screen for drug-drug and drug-disease interactions. Finally, the psychiatrist must help the patient and the treatment team understand the personal implications of the illness for the patient.

Social Security Disability (SSD) Versus Supplemental Security Income (SSI)

Both SSD and SSI require that you have a condition which prevents you from working, according to the rules and regulations of the Social Security Administration. To receive SSD you must have worked and paid Social Security taxes (FICA) for a sufficient period of time. SSD benefits are available regardless of your current financial situation and are based upon your past earnings. To qualify for SSI benefits, your household must have very limited income and resources. SSI benefits are available without regard to work history and are based on federal poverty guidelines.



Generally, SSD recipients become eligible for Medicare within 29 months of the onset of their disability, while in most states, SSI recipients become eligible for Medicaid either on the date they filed their application for disability benefits or their date of onset, whichever is later.

A disabled child typically will not have worked long enough (or at all) to be eligible for SSD, so children from lower-income families will usually only be eligible for disability benefits through SSI. In certain circumstances, adults may be eligible for both SSD and SSI benefits. Children, since they typically lack the work history required for SSD eligibility, will only be covered by SSI and only if their family meets income level requirements. Nevertheless, there are a few special situations in which a disabled child may receive SSD benefits.

Children under age 18, whether disabled or not, may be eligible to draw benefits on a parent's Social Security earnings record., if the parent is receiving disability or retirement benefits from Social Security or is deceased and the parent met insured status requirements. Children of an eligible worker may qualify for benefits as natural children, stepchildren, adopted children, and, in some cases, grandchildren. Benefit amounts depend upon a variety of factors.

A disabled child of an insured parent who is receiving disability or retirement benefits, or who has died, may be eligible for a benefit called Disabled Adult Child benefits (DAC) if the child is 18 or older and unmarried and if the disability began before age 22. This benefit may continue for the child's lifetime as long as the child remains disabled and unmarried, does not engage in substantial gainful activity resulting in benefit cessation, and does not become entitled to a higher benefit amount on another Social Security program. As with SSD, DAC benefits result in Medicare eligibility after being entitled to cash benefits for 24 months.

Support Group Leaders Meet for “First-

By KRISTA HALL and PAT DREIBELBIS

For the first time in its 18 years, the Charcot-Marie-Tooth Association gathered its support group leaders or their representatives in St. Louis, Missouri, to discuss the successes and failures of the support group concept and to strategize about ways to improve their own groups in the local areas. The meeting took place April 27, 28 and 29 at the Frontenac Hilton.

One of the leaders of the conference, Krista Hall, a registered nurse from New Market, Maryland, provided the narrative for this article. She began by commenting, “We saw Bob Budde and his wife Phyllis for breakfast on Friday morning. Phyllis is a registered nurse and has MS. Wow! What a team they make! I felt an immediate bond with the “nurse thing.”

“We set up a table in the lobby with the CMTA table cover on it and people stopped by as they checked in.”

The first official event, the reception on Friday night, was fun. Everyone was friendly and visited with everyone else. “The leaders were all talkative, but Rosemary Mills won the award in that department for the weekend (a good quality in a leader).” One of the other leaders, Ruth Levitan, of Berkeley, California, did an icebreaker where the attendees had papers and had to go around the room and ask people about various topics to find some points of commonality. Athena Diagnostics provided the food for the reception and “the toasted ravioli was a big hit.”

The program began with a personal story from Krista about her CMT. President Ardith Fetterolf spoke about her diagnosis of CMT and welcomed the attendees. Finally, Ann Beyer, Chairman of the Board, told about starting the



Dr. Florian Thomas, from St. Louis University, was the keynote speaker who helped attendees learn “How to be a successful CMT patient.”

first CMTA support group and what the CMTA can do for support groups and what the groups can do for the CMTA in return.

Krista then introduced Dr. Florian Thomas of St. Louis University, who presented a discussion entitled, “Being a successful CMT patient. How to get on your doctor’s nerves and feel good about it.” Dr. Thomas suggested asking your doctor how many CMT patients he sees on a regular basis. Three in the last twenty years isn’t a good sign! He also pointed out that you need to ask your orthopaedic surgeon how many foot and ankle procedures he does. You don’t want a back specialist working on your foot! His information was both informative and very “down to earth.” He took questions from the group and “everyone loved him.”

Bob Davidson, Community Relations Director for the St. Louis Hospital, spoke about Shriners’ services and gave out good maps of Shriners’ Hospitals and handouts of their programs. Following his presentation, the group broke for a “networking” lunch.

The session following lunch, called “Planning the Perfect Support Group,” centered around finding quality speakers, getting members to work on committees, meeting days and places and publicity for groups. The small groups who discussed these topics came back to the whole group and exchanged ideas. “There was excellent group energy and people were

Support group leaders and CMTA Board members gathered in St. Louis for the first national Support Group Leaders Conference.



Ever" National Conference

able to solve their own problems as they heard the various suggestions from others."

The final topic before the break for dinner was alternative or integrative medicine. The group discussed acupuncture, massage, reflexology and healing touch. Krista led a chakra or energy-centered lesson. The day ended with a self-relaxation exercise that the leaders could do for themselves. They were given copies of the exercise that they could take back to their groups. As Krista said, "I think most of them participated, but I had my eyes closed and only peeked once or twice."

A representative of the Neuropathy Association spoke to the group during dinner and had some handouts, including a good booklet on exercise and peripheral neuropathy.

Sunday morning began with a presentation from Larry and Donna Clark of Versailles, Missouri. There was a bit of humor involved in



Phyllis and Bob Budde and Marilyn Munn Strand enjoyed the Friday evening reception, where support group leaders got to know one another.

their introduction as Krista attempted to apply the French pronunciation to Versailles only to be told that in the Midwest they say Ver-sales. They had an excellent PowerPoint presentation on orthotics, AFOs, the boot they have helped to develop and how to get shoes to accommodate the CMT foot.

The weekend ended with a recounting of all the good things that happened during the weekend. To reinforce the energy of the conference, the meeting ended with a closing circle that produced some emotional responses. Then, it was off to the shuttle and the airport with the hope that the enthusiasm generated at the conference will follow each leader home and become the energy for new and better meetings at the local level.

Although I'm groggy from lack of sleep and jet lag, I'm excited about all I learned in St. Louis and the wonderful people I met. I'm in love with Ardith, Krista and Ann and all the other leaders. Role models all. Thanks for all you've done.

—Ruth

I would like to take this opportunity to thank you and your staff for all the effort that was involved in making this conference such a huge success. We all had a wonderful time and learned a great deal during our time together. Please express my sincere thanks to all who worked very hard in putting this event together.

—Bill



Krista Hall, one of the organizers of the support group conference, did a presentation on alternative healing methods such as massage and self-relaxation methods.

Support Group Leader Angela Piersimoni Takes the CMTA's Work to the Local Level

I have been asked to introduce myself to you as the support group leader for the Horseheads, New York, regional CMT group and to share a bit of my experience with the diagnosis of CMT.

In late summer, 1985, I awoke one morning with a tingling sensation which extended down my arms and my entire chest area, sternum to lower ribs. Because I had been a medical secretary for a number of years prior to that day, I quietly went through the list of disease processes that might or might not be responsible, i.e. heart attack, ulcer, anxiety. My mother was living with us at that time and her health was failing. So, I thought I should get this checked out promptly.

Almost two years later, after many tests, including esophagoscopy, a trial of hand splints and neck collar, MRI, bone scan, myelogram,

and of course, EMG/nerve conduction velocity testing, my neurologist came up with a tentative diagnosis of CMT. Following this, I was referred to the Neuromuscular Clinic of Strong Memorial Hospital in Rochester, NY. Dr. Kingston, the clinic neurologist, asked for a sural nerve biopsy for confirmation. Following this testing, I was "officially" diagnosed.

Truthfully, I can't recall (with any certainty) how or when I became aware of the Charcot-Marie-Tooth Association. I believe my MDA representative from the Binghamton, NY, office informed me about it. When the correspondence from the CMTA arrived, it contained the question, "Would you wish to start a support group?" Thinking it would be great to have some support from others involved with the disease, I decided to give it a try. With much help from Pat Dreibelbis at the CMTA office, who sent out the first announcements, the Horseheads regional support group was begun.

On the night of the first meeting, my husband, Joe, and I waited expectantly for people to arrive. Many people came into the building that evening, but all but two of them were headed to the monthly meeting of the Audubon Society being held in the room next to ours! Disappointed, but nonetheless determined, we continued and today the group consists of approximately 15 members. Our active list waxes and wanes for many reasons; however, each time we receive a call from a person who has no support group nearby, I realize how important support groups are. I am pleased to listen and let them know we understand their concerns and problems. These people with CMT are called our extended support group.

(Editor's note: Angela did not "brag" about her husband, Joe, but he has been her assistant in running the group since its inception. He even attended the national support group leaders' conference and participated in the break-out sessions. A supportive family member is one of the most significant characteristics of a successful support group leader!)



A supportive family is a tremendous help to support group leaders. Here, Angela Piersimoni (far left) posed with her family members, grandchildren Jessica and Josh, and son Eric and his wife Linda. (Her husband, Joe, is taking the picture.)

CMTA Support Groups

Alabama/Greater Tennessee Valley

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

Arkansas—Northwest Area

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

California—Berkeley Area

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

California—Los Angeles Area

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

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

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

Colorado—Denver Area

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

Florida—Boca Raton to Melbourne

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

Florida—Miami/Ft. Lauderdale

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

Kentucky/Southern Indiana/ Southern Ohio

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

Massachusetts—Boston Area

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

Michigan—Detroit Area

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

Michigan—Flint

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

Minnesota—Benson

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

Mississippi/Louisiana

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

Missouri/Eastern Kansas

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

Missouri—St. Louis Area

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

New York—Greater New York

Place: NYU Medical Center/
Rusk Institute, 400 E. 34th St.
Meeting: Monthly
Contact: Dr. David Younger,
212-535-4314, Fax 212-535-6392

New York—Horseheads

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

New York (Westchester County)/ Connecticut (Fairfield)

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

North Carolina—Archdale/Triad

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

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

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

Ohio—Greenville

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

Oregon/Pacific NW

Place: Portland, Legacy Good Sam
Hospital, odd months
Brooks, Assembly of God
Church, even months
Meeting: 3rd Saturday of the month
(except June and Dec.)
Contact: Jeanie Porter, 503-591-9412
Darlene Weston, 503-245-8444
E-mail: jeanie421@yahoo.com or
bizerbabe@aol.com

Pennsylvania—Philadelphia Area

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



Members of the CMTA's Medical Advisory Board answer questions from readers.

Ask the Doctor

(Editor's note: All of this issue's questions were posed at the national support group leaders' meeting and were answered by Medical Advisory Board member, Dr. Florian Thomas.)

Dear Doctor,

How much are internal organs affected by CMT 2? What effect do hot and cold and altitude have on CMT patients? Could they affect a rapid progression?

The doctor replies:

In general, CMT1 and CMT2 affect only nerves supplying the skin and muscles that are under our conscious control, but not nerves supplying internal organs or blood vessels. There are very rare families in which, for instance, diabetes or lipid abnormalities are associated with CMT. In general, an extensive search for causes unrelated to CMT must be completed for any medical problem that a CMT patient may suffer from before CMT is suspected as the cause of that problem.

Heat, cold, barometric pressure and altitude do, in general, not affect symptoms in any specific way; however, if a CMT patient flies, let's say from Detroit to Aspen, he or she may feel weaker due to lower oxygen pressure, as may anybody without CMT. Sensory symptoms such as tingling or burning are often affected by temperature changes, but should return to baseline when the temperature returns to what the patient is used to. Of course, frost bite could damage skin and cause discomfort in CMT patients as in anyone else. But otherwise, extreme heat or cold should not trigger disease progression. Any instance of disease progression that is more rapid than expected warrants an extensive search for causes.

Dear Doctor:

I was told that CMT had a stocking and glove effect, but now I notice that I have numb spots and tremors in other parts of my body which I think might be related. Can you comment?

The doctor replies:

While stocking and glove is the normal distribution of sensory changes, there is another condition, HNPP, in which the nerves are exquisitely sensitive to compression, entrapment, and trauma. This could produce patchy weakness and numbness. Tremors, on the other hand, are common in CMT. Certain forms of tremor are commonly found in the general pop-

ulation without CMT and may occur in CMT patients just by chance, while others are often associated with neuropathies.

Dear Doctor:

What diseases are commonly co-morbid with CMT?

The doctor replies:

Very few, and typically, none. Rarely there is evidence of brain or spinal cord involvement or diabetes and this has been categorized as CMT with additional features or atypical CMT. Most of the time, when such additional signs are present, they are not due to CMT, but result from an unrelated disorder, and need appropriate independent investigation. Cardiac abnormalities have been described as well as cases associated with myotonic dystrophy. Several CMT MAB members are currently working on speech and voice (laryngeal) and breathing (phrenic) disorder associations in CMT. Most of the time, when such additional signs are present, they are not due to CMT but result from an unrelated disorder.

Dear Doctor,

I have recurring, pulsating electrical shock-type pain in various areas of my feet. It occurs about 3-4 times a month. It lasts at least 24 hours with a zapping pain every 30-60 seconds. I cannot sleep during this time period. Of course, I am exhausted afterwards. I take amitriptyline (25 mg at night), which has helped a little.

The doctor replies:

None of my CMT patients has mentioned this complaint, but a similar complaint was discussed earlier this year in the CMTA newsletter. It certainly sounds like a pain originating in a nerve. Nerve root compression in the back must be considered as an alternative explanation. One hates to give daily medication for a pain that occurs 3-4 times a month, but the kind of medication that works for this must be taken chronically. Amitriptyline is a good drug but is rather sedative and has to be used with caution in older people. Several of its cousins, nortriptyline and desipramine, are less sedative. If well tolerated, better pain relief might result from a higher dose. They are all convenient because they are taken once a day. Gabapentin has fewer side effects but is usually taken 3 times a day. One other option might be a TENS unit, which has electrodes placed on the affected limb through

which a small electric current flows from a battery. This often alleviates this kind of pain.

Dear Doctor,

I am very frightened by the use of prescription pain medication and would like to know what is best to take for the pain associated with CMT. Does the pain ever stop or is it just one of those lifelong things we must endure?

The doctor replies:

While it is understandable that someone wants to limit the medications he or she takes, chronic pain is dangerous in itself. It can lead to great stress, poor sleep patterns and depression. Most importantly, the cause of pain must be determined. CMT patients are prone to orthopaedic pain in their feet and other joints, including the back, and this pain must be approached differently from neuropathic pain or tingling pain. Ankle braces and other orthotic devices can alleviate certain forms of pain, though sometimes surgery is needed. For tingling pain, drugs that are otherwise used to treat seizures or depression are often highly effective.

Whether pain stops is dependent on the type of pain. If it results from foot deformities, it may be stopped by surgery. Tingling pain can often be stopped by medications or it can just resolve on its own.

Dear Doctor,

A five-year-old boy suspected of having CMT has a cyst on his wrist. Could there be any connection with the disease and this cyst?

The doctor replies:

I cannot answer this definitively, though no association comes to mind. There are many kinds of cysts, with different causes. I would suggest you ask his pediatrician.

Dear Doctor,

Can coenzyme Q-10 or Omega 3 help strengthen my weakened muscles?

The doctor replies:

While Coenzyme Q-10 may have a role in some neurological conditions and is being investigated, I am not aware of a role for this compound in CMT. Similarly, I am not aware of any studies published in reputable journals showing a benefit for Omega 3 supplements.

In general, if a compound is safe, not prohibitively expensive and appears to help, I am not going to dissuade a person from taking it. However, over the counter compounds such as CoQ-10 and Omega 3 do not undergo the rigorous efficacy and safety testing that FDA drugs do, and there are theoretical concerns that they might have negative side effects. This is why patients must be very cautious about them.

Dear Doctor,

I did some research on the Internet for dry, brittle nails because I've been experiencing that and I found out that it can be a symptom of hypo- or hyperthyroidism. I thought I remembered hearing that people with CMT should be tested for thyroid problems and I wondered if we are at risk for these conditions. If so, why would we be?

The doctor replies:

Dry, brittle nails can be a feature of many different conditions. In general, searching for a symptom on the Internet can lead to all kinds of conclusions which may or may not be correct. So, by only looking at thyroid disorders, a patient and his/her doctor might miss an important alternative explanation.

Patients with neuropathy (CMT or other) must be checked for several underlying conditions that may be the primary or an additional cause for some of their problems. 99% of all CMT patients are not at higher risk for thyroid diseases. But regardless, I check CMT patients for thyroid disorders, diabetes, syphilis, B12 deficiency, rheumatoid diseases, anemia, liver and kidney disease and nerve compression. The goal is to find any condition that might make CMT worse and could be corrected.

Dear Doctor,

Could marijuana make CMT worse?

The doctor replies:

The effects of marijuana on nerves have not been well studied. It is still illegal in this country and modern-day marijuana appears to be more potent than the stuff of yesteryear. It does appear to have a toxic effect on the brain, but whether this is less or more than alcohol used at a similarly mind-altering dose is not clear. If a person falls asleep in any intoxicated state and compresses a nerve, this can cause damage. Such damage might be worse in CMT patients.

GIFTS WERE MADE TO THE CMTA

IN MEMORY OF:

Dorothy Blanchard
Kay Flynn

Richard Fernandes
Mike & Pam Gorman

Ralph Hall
Arthur & Kathleen
Benedict

John Hill
Jeanne Andriot
Kenneth Freitag
Gerrit & Janet Gucky
Richard & Mary Hansen
Richard Hunter, Jr.
Anne Janzer-Pfeil
Thomas & Milou Kelley
Vic & Caroline Knowlton
Kimberly Merino
Mr. & Mrs. Warren
Rasmussen
Mr. & Mrs. John Russell
Wisconsin Electric
Power Company

IN HONOR OF:

Gina & Stephanie DiCara
Arden Kolb-DeBolt

Mr. & Mrs. Joe Gelman
Leon Gelman

Neal Gossard
Judy Gossard

Megan Knuth
Dwain & Peggy Knuth

James Moneyhon
Caroline & Mary Jean
Shugars

Eric & Michael Ponder
Judy Gossard

Rachel Weil
Leon Shapiro

Hart Wurzburg
Virginia Sonnenschein

Professionals Working for CMT Patients: Dr. Florian Patrick Thomas

(Editor's Note: Dr. Thomas has been a member of the Medical Advisory Board of the CMTA since 1998. His involvement in this April's National Support Group Leaders' conference is just one of the examples of his intimate involvement in patient care and education.)

Dr. Florian Thomas calls St. Louis, Missouri, his home now, but his trip to St. Louis University has been a long and impressive one. He was born in Germany and educated at the University of Bonn, receiving his B.S., his M.A in psychology, and his M.D. there. During his time at the University, he received clinical training at Harvard and Case Western Reserve and later at Columbia and McGill.. He received his Ph.D. in retrovirology and molecular biology from McGill University in Montreal in 1995. Dr. Thomas has been involved in neuromuscular research for over 15 years. He runs the neuromuscular center at the St. Louis VA Hospital and at Saint Louis University, where he is on the faculty as an associate professor of neurology, and sees many patients with CMT.

However, it is the personal interest that Dr. Thomas has taken in Charcot-Marie-Tooth patients that truly makes him outstanding. In 1997, he helped develop the St. Louis Support Group of this organization and the St. Louis chapter of the Neuropathy Association. His work with a large family of CMT patients in the St. Louis area and elsewhere in the country was reported on in a previous issue of the newsletter. Just this April, Dr. Thomas was the keynote speaker at the National Support Group Leaders' meeting in St. Louis. He told the audience, "When I met Carole Haislip (co-leader with him of the support group), I had to look at 65 other people in her family. She jump-started my CMT patient base."

His presentation that day, entitled, "Being a successful CMT patient: How to get on your doctor's nerves and feel good about it," indicates just how much he advocates that patients demand the best care from their various specialists. Before choosing a doctor or specialist, he urges patients to ask how many CMT patients the doctor sees. Having a stroke



Dr. Florian Thomas serves on the CMTA's Medical Advisory Board and co-chairs the St. Louis support group.

specialist treat your CMT is not nearly as comforting as having a person who treats the various muscular dystrophies.

He pointed out that doctors often think they can, or should be able to, do everything for their patient. In actuality, patients need to know that one doctor can't do everything and should be willing to refer them to a physical therapist, orthotist, surgeon or whomever is needed. Communication back from the specialist to the primary care doctor is essential. The goal should not be to make a diagnosis and be done with it. The relationship needs to be ongoing. Continuity is important so that changes are noticed and a doctor can recognize when something else is going on. As Dr. Thomas said in his presentation, "In Germany, they say you can have both fleas and lice" so it's important to notice changes that indicate a second problem which needs to, and can be, fixed.

Dr. Florian Thomas combines the best of both worlds—an academic and clinical excellence with humanity and an empathy for each patient that he sees.

The CMTA Report

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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.

Letters to the Editor:

Dear CMTA,

I am a 45-year-old woman diagnosed with peripheral neuropathy. It has been over three years now and it gets worse every year. I have chronic pain in my feet and numbness up my legs with weak ankles. My hands hurt and tingle all the time, and I get pain in my wrists and up to my elbows. I wear gloves for gripping and braces for my wrists. I have a brace for my left leg, and I use a cane now for my balance.

I try to understand this disorder, but it is hard at times. I have good days and bad days. The pain in my feet keeps me from doing much. I am taking Neurontin and Vicodin for pain.

I have joined the CMTA and the news reports I get are helpful. I really enjoy getting your reports and learning all that I can. I hope to be able to attend a support group some day. Thank you for all your good work and all the research.

—Chris C., MA



Dear CMTA,

I am a physical therapist and a patient with a recent diagnosis of CMT. I am 46 years old and the mother of 4 children. At age 44, months after being diagnosed, I agreed to participate in a

sural nerve biopsy for research in the development of treatment for CMT. The aftermath has been confusing to say the least, and I have been told that much of my lack of full recovery is very unusual. The progression of my CMT was more extensive after the biopsy report, and I wonder if there is a correlation between surgery on patients with a pre-existing neuropathy as opposed to a patient with no neuropathy. The symptoms now for almost 2 years are a dull ache at the 2-inch incision site on the posterior calf (5-7 inches from the floor) and an electrical needles feeling in the foot which can be quiet but becomes active with increased physical activity. The other disturbance is the large area of complete numbness in my lower calf, ankle and heel and 1/3 of the foot itself. I may be different, but maybe I am not alone.

There were at least 25 other participants in this study. I would like to hear from any post-sural nerve biopsy patients, research related or other, but especially if the incision is the higher cut, as is mine (some are done below the ankle). If anyone can shed some light on my experience by sharing theirs, I would be grateful.

—Pam Lett
3762 Quail Hollow Drive
Columbus, OH 43228



WRITE TO US!

Pat Dreibelbis, Editor

The CMTA Report

CMTA

2700 Chestnut Pkwy.

Chester, PA 19013

The CMTA reserves the right to edit letters for space.

CMTA Remembrances

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

Honorary Gift:

In honor of (person you wish to honor)

Send acknowledgment to:

Name: _____

Address: _____

Occasion (if desired):

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

Memorial Gift:

In memory of (name of deceased)

Send acknowledgment to:

Name: _____

Address: _____

Amount Enclosed: _____

- Check Enclosed
- VISA
- MasterCard

Card # _____

Exp. Date _____

Signature _____

Gift Given By:

Name: _____

Address: _____



MEDICAL ALERT:

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

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

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

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



What is CMT?

- ... is the most common inherited neuropathy, affecting approximately 150,000 Americans.
- ... may become worse if certain neurotoxic drugs are taken.
- ... can vary greatly in severity, even within the same family.
- ... can, in rare instances, cause severe disability.
- ... is also known as peroneal muscular atrophy and hereditary motor sensory neuropathy.
- ... is slowly progressive, causing deterioration of peripheral nerves that control sensory information and muscle function of the foot/lower leg and hand/forearm.
- ... causes degeneration of peroneal muscles (located on the front of the leg below the knee).
- ... causes foot-drop walking gait, foot bone abnormalities, high arches and hammer toes, problems with balance, problems with hand function, occasional lower leg and forearm muscle cramping, loss of some normal reflexes, and scoliosis (curvature of the spine).
- ... does not affect life expectancy.
- ... has no effective treatment, although physical therapy, occupational therapy and moderate physical activity are beneficial.
- ... is sometimes surgically treated.
- ... is usually inherited in an autosomal dominant pattern, which means if one parent has CMT, there is a 50% chance of passing it on to each child.
- ... Types 1A, 1B, 1X, HNPP and EGR-2 can now be diagnosed by a blood test.
- ... is the focus of significant genetic research, bringing us closer to answering the CMT enigma.

The CMTA Report

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



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