

4

Tribute to
Dr. Robert
Lovelace

8

Cycling to
Raise Money
for Research

11

April 5th
Conference
Review

14

Pros and
Cons of
DNA Testing

The CMTA Report

SPRING 1997

Vol. 12, No. 2 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.

Kay Flynn Receives 1997 Rebecca Sand Volunteer of the Year Award

Non-profit organizations not only depend on volunteers; they cannot exist without them. The CMTA has benefitted from incredible volunteer support. We are grateful for the ongoing commitment of countless individuals who devote their time and resources to the CMTA. Rare individuals distinguish themselves through their service and, in so doing, garner the interest and involvement of others. To acknowledge our debt of gratitude and to highlight the accomplishments of these leaders, the CMTA annually recognizes a Volunteer of the Year. Kay Flynn, of the Westchester Support Group, is the recipient of this year's award, which was deservingly named in honor of Rebecca Sand.

Though she died in 1995, Rebecca's name and efforts are well known to many. Her "pleasant persistence" will long be remembered. Whether she was cajoling people to get involved or persuading them to contribute, she was a valuable advocate for the organization. Rebecca's friends and relatives frequently honor her memory and carry on her tradition of support through their own generosity and service. Though the Volunteer of the Year is chosen annually, this recognition sometimes reflects an "outstanding year," but more often, it reflects long and dedicated service. This is certainly true of Kay Flynn and past honorees: Bob Daino, Dr. Gareth Parry, Sue Tarpinian, and of course, Rebecca Sand.

Kay Flynn richly deserves the 1997 Volunteer of the Year Award for a truly great year this year and for many years past. She and her support group members came to the Yale Conference in October to help make that a success and have organized several important meetings of



Kay Flynn (left) purchased a baby quilt from Gloria Charny, who, in turn, made a contribution to the CMTA. In addition to Gloria's fundraising initiative, the Westchester Support Group will undertake other fundraising efforts to support the CMTA.

CMTA BOARD

Ed Butchko
Chairman

Diane Freaney
President

Ann Lee Beyer
Vice-President

Jack Walfish
Treasurer

Ardith Fetterolf
Corresponding Secretary

Krista Hall
Recording Secretary

Robert E. Lovelace, M.D.
Chairman, Medical
Advisory Board

Joe Beernink
David Grachek
Steven E. Khosrova
Loren Miller
Michael Molinari
William Quinn, DPM
Louise Smith
Rodman Steele, Jr.

VOLUNTEER OF THE YEAR RECIPIENTS

1992	Rebecca Sand
1993	Sue Tarpinian
1994	Bob Daino
1995	Dr. Gareth Parry
1997	Kay Flynn

continued on page 3

Volunteers Give Life to the CMTA

In the world of small non-profit organizations, it is by the strength of its volunteers that an organization grows and succeeds in its ambitious goals. Three groups, the Board of Directors, the Medical Advisory Board, and the CMTA's support groups are largely responsible for the success to date of the CMTA. In this newsletter, we recognize and thank each of those groups for the work they do, freely and energetically, for all of the members of this organization.

CMTA Board of Directors Meets in February

The Board of Directors of the CMTA comprises 17 people from all over the United States who work in every professional field imaginable and who share one passion: to make the CMTA be a recognized world leader in promoting awareness of Charcot-Marie-Tooth Disorders and in developing solutions for the effects of CMT now and for the future. With that common goal in mind, the Board met in Princeton, New Jersey on February 8th and 9th, to examine where the organization is in relation to its goals and vision and where it could go in the next few years.

One of the most important topics of discussion was the creation of a mission statement that would provide direction for the work of the organization. The final version of the CMTA's mission statement was, "To create awareness of Charcot-Marie-Tooth (CMT) disorders within the health care community and the general public, and to be a leading source of information regarding CMT disorders. To

encourage, promote and support research into the causes, treatments, and cure of CMT and to facilitate education and support for persons affected by CMT."

Another major topic of discussion was the Third International Research Symposium on Charcot-Marie-Tooth Disorders, which is tentatively planned for Montreal, Canada in October of 1998. This conference will bring together noted CMT researchers from all over the world who will present papers and share information regarding both genetic and therapeutic research findings. The Third International Conference is a huge undertaking, involving hundreds of researchers and clinicians, and will require a budget larger than any previous commitment made by the CMTA. Dr. Michael Shy of Wayne State University and Dr. Robert Lovelace, Chairman of the Medical Advisory Board, are organizing the scientific presentations for this conference.

The 17 members of the Board of Directors not only gave up a weekend in the winter to meet, but they also meet in person or by telephone conference call every other month. In addition to meetings, they organize and attend regional patient/family conferences, man the CMTA booth at conventions of neurologists, podiatrists, physiatrists, and physical therapists, run and/or attend local support groups, write fund-raising letters, manage the finances of the organization, set up and maintain the Charcot-Marie-Tooth Association web site, host fund-raising events in their home states, and a million other day-to-day tasks that keep the CMTA vital and growing...and they do all of this as volunteers!

Attendees at the Board Retreat in Princeton, NJ: (seated, left to right) Bill Quinn, Rod Steele, Paul Flynn, Joe Beernink; (standing, second row) Diane Freaney, Jack Walfish, Ann Beyer, Ardith Fetterolf, Krista Hall; (standing, third row) Steve Khosrova, Ed Butchko, David Grachek, Pat Dreibelbis.



Kay Flynn Volunteer of the Year
continued from page 1



their own. The group hosted Dr. Jesse Cedarbaum, Vice President, Clinical Affairs of Regeneron (a bio-tech company whose research has implications for people with CMT) and recently heard a talk on “magnetic therapy.” In addition to their cutting-edge presentations, part of what makes Kay’s group so special is the strong sense of camaraderie. Beyond sharing their personal experiences with CMT, Kay asks members to keep one another informed about other matters of interest such as products and adaptive devices, types of shoes, doctor or specialist referrals, Social Security and Medicare, and women’s health issues. Members also commonly exchange letters and cards for birthdays, anniversaries, or get-well wishes.

It was not always bustling activity and well-attended meetings. Mary Rehm, Kay’s sister and right-hand in leading the support group, recalls their humble beginnings. Mary said, “We had put our meeting notice in a local paper. Our “kick-off” was me, Kay, and our sister Alfreda (Gomez) and a couple visiting from France who saw our ad. We’ve grown to nearly 40 people in 8 years, but that French couple never did come back!” The group collects dues to cover expenses and is considering some creative fundraising ideas to support the CMTA. Kay purchased a baby quilt made by Gloria Charny, who has donated the money to the organization. (See photo, page 1.)

There are dynamic and thoughtful people working with Kay in the Westchester Support Group. One is Dan Iacovella, who had this to say about the Rebecca Sand Award recipient: “What has always amazed me about Kay is that she seems to remember the different things that are important to each person in the group and integrates those things at the appropriate time so that everyone feels like they belong. This is just one characteristic that exemplifies her selfless dedication to people with CMT. She has always made helping us and those in the larger CMT family her first priority, above her own needs. This is what makes her so wonderful and why we are so thankful. We are spoiled to have a person with tremendous energy leading our group and privileged to have a person of such integrity in our lives.”

Kay, the CMTA agrees wholeheartedly. Congratulations and many thanks for all you do!

CMTA MEMBERSHIP/ORDER FORM

Name: _____

Address: _____

Phone Number: _____

	QTY	COST	TOTAL
Charcot-Marie-Tooth Disorders: A Handbook for Primary Care Physicians		<i>members</i> \$15 <i>non-members</i> \$20	
Membership Dues		\$25	
CMT Facts I <input type="checkbox"/> English <input type="checkbox"/> Spanish		\$3	
CMT Facts II <input type="checkbox"/> English <input type="checkbox"/> Spanish		\$5	
CMT Facts III		\$5	
VCR Tape: CMT Neurology		\$15	
VCR Tape: Physical Therapy & Occupational Therapy		\$15	
VCR Double Tape: duPont Conference		<i>members</i> \$25 <i>non-members</i> \$40	
CMT Informational Brochure <input type="checkbox"/> English <input type="checkbox"/> Spanish		FREE	<i>Send Self- Addressed Stamped Envelope</i>
Physician Referral List: States: _____		FREE	
Letter to Medical Professional with Drug List		FREE	
Contribution to CMT Research			
TOTAL			

Checks payable to the CMTA

VISA MasterCard

Card Number _____ Expiration Date _____

Signature _____

Mail to the CMTA, 601 Upland Ave., Upland, PA 19015

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.

A Tribute to a Volunteer Leader: Dr. Robert Lovelace

Dr. Robert Lovelace, M.D., F.R.C.P., chaired the meeting of the CMTA's Medical Advisory Board during the American Academy of Neurology Meetings in Boston this past April.



There are many people who have been responsible for the growth, success, and recognition of the Charcot-Marie-Tooth Association over the years, but none whose association with the organization has been as long-lasting or as impact-filled as that of Robert Lovelace, MD, FRCP. Dr. Lovelace has been a medical advisor to this organization since 1984, when he was asked to expand his advisory position from the local New York City MDA Charcot-Marie-Tooth Support Group to the national organization.

In 1984, he was the only doctor functioning as a medical advisor to the CMTA (then called the National Foundation for Peroneal Muscular Atrophy). By 1987, while planning the Second International Conference on Charcot-Marie-Tooth Disorders, he had formulated the idea of a medical advisory board and begun recruiting members. Today, approximately 49 internationally renowned clinicians and researchers serve the CMTA with their medical and scientific expertise and Dr. Lovelace has been the chairman of that group since its inception.

Dr. Lovelace's interest in Charcot-Marie-Tooth Disorders began back in the early

1960s when he was a fellow at the Mayo Clinic in Rochester, Minnesota. There, he noticed conduction abnormalities in patients with Charcot-Marie-Tooth Disorders and when he went to Columbia in 1963, he began collecting cases to substantiate the published theories of Dr. Peter Dyck and Dr. Edward Lambert on the various types of CMT.

Among Dr. Lovelace's contributions to the CMTA are his attendance at all meetings of the Board of Directors, his chairing of the semi-annual meetings of the Medical Advisory Board of the CMTA, his review of post-doctoral fellowship grant applications, his answering of endless "Dear Doctor" questions for this newsletter, and, of course, his encouragement for the writing and publication of *Charcot-Marie-Tooth Disorders: A Handbook for Primary Care Physicians*.

In addition to his volunteer work for the Charcot-Marie-Tooth Association, Dr. Lovelace also serves on the medical board of the Myasthenia Gravis Foundation of America and continues an active practice at the Neurological Institute of Columbia Presbyterian in New York City.

When asked about his long and successful association with the CMTA, Dr. Lovelace said, "I've been lucky to have had a good working relationship with each of the lay leaders of the CMTA through the years. That's key to the success of an organization and its medical advisors."

The CMTA has also been "lucky" to have had Dr. Robert Lovelace as the dedicated Chairman of its Medical Advisory Board.

GIFTS WERE MADE TO THE CMTA IN MEMORY OF:

Anne Bernstein
William Bernstein

Louise R. Bessey
MaryAlice V. Ralls

Merrill Bliss Park
George & Maxine Hardy
Fred Rach
Margaret & William Sowle

Frances Boyd
Edward & Pauline Mulvaney

Vanoy Bradford
Janice Rensing

Samuel C. Cantor
Eddy & Helaine Cantor

Jennie Campanella
Farrell Insurance Agency, Inc.:

Gail Bortolotti
Andrea Leonard
JoAnn Tripp
Sue Provencher
Sandy Leonard
Grace Levinson
Joan Grubbs
Brenda Viera
Raylene McSully
Lori Paluzzi
Carrie Moniz

Catherine Arecchi
Catherine Campanella
Mr. & Mrs. Frank Chiaretta

Dr. & Mrs. John Demman
Mr. & Mrs. Richard Fumanti
Susan Leto
Lucy Lopardo
Mr. & Mrs. Carlo P. Martorana
Carlo P. Martorana
Louis Massiano
Ralph & Teresa Maurizi
Joseph McLaughlin
Jane T. Melone
Joan O'Sullivan
Printronic
Maria C. Winkelmann

Nellie Nan Charles
Ed & Apryl Chidiac

Support Groups: They Can Really Work

The Kansas City support group meets once a month at the Mid-America Rehabilitation Hospital in Overland Park, KS, a suburb of Kansas City. Members of the group live in and around Kansas City, coming from as far as Topeka, Paola, Baxter Springs, and Lawrence, KS, and Blue Springs, MO. Meetings are on the first Saturday of the month in the hospital cafeteria, with drinks and breakfast snacks furnished by the hospital.

One member of the group has the meeting notices printed and mailed by his company and his company secretary monitors voice mail and keeps a mailing list of all 51 members.

This self-help group started 5 years ago with three members, including the founder, a young college student. She remains a primary driving force in the group, helping to organize the new St. Louis support group and also making the initial contact that resulted in the family conference at Shriners Hospital in St. Louis.

The diverse group also includes:

- A retired airline purchasing agent and his wife, who are involved in all sorts of volunteer projects but have taken the time to assist several members with Social Security disability applications. This couple has also traveled to CMT family conferences in several states.
- A talented fiber weaver who has assembled a CMT library available to all members of the group. The library includes VCRs, past CMTA newsletters and anything else pertaining to CMT. Her husband, a US magistrate, attends the support meetings and lends his legal expertise when needed.
- A college professor, a CMTA Board Member, and a machinist.

- A couple who do the PR work for the group and whose efforts have resulted in many new members joining the group.
- A three-generation family with CMT who are originally from Australia.
- A business owner who found he had CMT in later years, and whose contributions enable the group to remain dues free.

Several members are unable to work but are always there to help with calling or finding speakers for the meetings. Many members have full-time employment and cope daily with CMT. One member helps her granddaughter by taking her on trips to Shriners Hospital for treatment.

Fifteen members of this group attended the St. Louis Conference and helped plan the Friday evening hospitality reception as well as tending to all sorts of chores on Saturday at the conference.

One reason that the Kansas City group has been able to grow and serve its members so well is because they are a diverse and happy group who care deeply for one another.

These members of the Kansas City support group of the CMTA (sitting: Lester and Winifred Johnson, standing: Lee Ann Borberg) helped organize and staff the St. Louis Conference Hospitality Suite.



The next newsletter will feature an updated listing of CMTA support groups by region, location, date and time of meetings, and the telephone number of the support group contact person. Currently, support groups are being surveyed as to those details. If you have not responded yet and are currently holding support group meetings, please return the forms to the office or your group will not be listed in the summer newsletter.

Richard L. Claflin

Nancy & Sheila Ballard
Environmental Research Institute of Michigan (ERIM)
Friends in the Accounting Department @ ERIM
Fred & Myra Gottlob
Linda & John Gunnis & Family
Mr. & Mrs. R. C. Halseth
Mr. & Mrs. Ray Hawks
Mr. & Mrs. Richard Hemp
Ms. Helen Holstein
Mr. & Mrs. Joseph C. Littmann

C. O. Dahlgren

Etta B. Dahlgren

Jeffrey A. Daily

David & Julie Arnold
Jennifer Ball & Ellen M. Dissauer
Geauga County Dept. of Human Services
Tom Grabarczyk of Labor Relations Mgmt., Inc.
Susan Larson for the Lake Lucerne Board of Trustees
Ernest & Teri Malnar
Northwest Tri-County Intermediate Unit
Mr. & Mrs. Wilton K. Town

Ellen Epstein

Marvin & Adele Greenwald

Irving J. Finkelman

Nancy & Howard Finkelman

Ben Fiscella

Kay Flynn
Peg Leahy

Iris Golmitz

Harriet Hartle

Maude A. Griner

Velma L. Griner & Virginia L. Wetcher

Zelda Herman

Bev & Bob Warner

Florence Hertzberg

Nancy Correia

Mildred Kesterson

June & Frank Gunnison

Dr. David P. Leahy

Kay Flynn
Florence Fiscella & Family

George Lowney

Sharon Lowney

Josephine Mondo

Dr. & Mrs. James Trezza

George F. Phy

Elizabeth Oliver

Rebecca Sand

Rhoda & Stephen Sand

Paul J. Terrell

Mr. & Mrs. William L. Pastor

Marguerite Zambito

Eleanor Z. Rabin

GIFTS WERE MADE TO THE CMTA IN HONOR OF:

Mrs. Brucie Rowe
Clay G. Battle

Cathy Brooks
Jan Dunham

James L. Emerson
Dr. Sean D. Royer

Cyd Kelly
Charisse K. Kelly

Rex Morgan, Jr.
C. Hilyard Barr
Luella H. Morgan

**Dr. Shirley Moyer's
Birthday**
Alessandra & Jim Trezza

**Mr. & Mrs. Frank
Williams' Anniversary**
Alessandra & Jim Trezza

**Sara Wainwright's
7th Birthday**
Barbara Cook

Medical Advisory Board Meets in Boston

The CMTA Medical Advisory Board (MAB) met in Boston on April 15th during the Annual Meeting of the American Academy of Neurology. Twenty MAB members took time out of their busy schedules to share their research and discuss new developments in CMT. These dedicated professionals meet twice a year at professional meetings. They are in touch during the year to collaborate on issues and keep informed on progress in the field of CMT research.

Dr. Jeffrey Vance and Dr. Michael Shy discussed the research conducted in their labs that is or has been funded by the CMTA. Dr. Vance's colleague, Dr. Peter Denton, was awarded the Anita Harding Fellowship in January 1996. His work involved looking for the gene that causes one form of CMT2. Using DNA gathered from many families with CMT, he localized one form of CMT2, called CMT2A to the very top of chromosome 1. This gene causes CMT in some families with type 2, but not all. The work on type 2 CMT is continuing in Dr. Vance's laboratory.

Dr. Agnes Jani received the first Armington Fellowship in January of 1997. Dr. Shy reported that Dr. Jani is continuing work that was begun last year. She is developing viral vectors as a way to introduce genes into Schwann cells. She's working on the delivery system to try to prolong

the expression of a gene by immune suppression in the animals prior to introducing the adenovirus into their sciatic nerve.

The members of the summer fellowship review committee for this year's fellowships were Dr. Thomas Bird, Dr. Kenneth Fischbeck, Dr. Roger Lebo, and Dr. Gareth Parry. The CMTA research community is small, with a majority of the major researchers included as MAB members. For this reason, fellowship committee members are chosen from the MAB. However, MAB members with fellowship applicants who are working in their labs are excluded from serving on the fellowship committee for that round of awards.

The committee reported that they had many excellent applications and found it difficult to make a decision. The CMTA had planned to award three grants for this summer, but was persuaded by the MAB committee to award four. These four grant awards are discussed in another article in the newsletter.

Another major topic of discussion was the Third International Research Symposium on Charcot-Marie-Tooth Disorders planned for October 1998 in Montreal, Canada. Dr. Michael Shy has been appointed Chairman of the Program Committee for the conference. Dr. Lisa

The Caligamed Ankle Brace: A Review

By **William Quinn, DPM, FACFAS**

The Caligamed Brace is an innovative, new support system that comes in both right-footed and left-footed versions, in various sizes. The braces were originally designed to support a healing or sprained ankle; however, it has excellent medial and lateral stability along with support of the foot in dorsiflexion.

The brace is light-weight and very easy to wear. It would act more as a temporary brace than a long-time brace. Many CMT patients need a full lower-leg AFO custom-fitted owing to the severity of their CMT and their atrophy. Some CMT patients require no long leg bracing at all.

I feel the Caligamed Brace is an excellent substitute for the patients in the middle group who require some support of their drop foot for toe clearance and some medial and lateral support to help prevent ankle sprains. The brace is relatively inexpensive when compared with custom-fitted AFOs and is extremely durable. It should last a long time. The brace has a low profile and can be worn either over or under socks. It will fit into most commercially bought shoes.

As with any bracing for support of patients with CMT, a diagnostic work-up and recommendation from an appropriate physician or



Dr. Betty Soliven, Dr. Thomas Bird, and Dr. Gareth Parry gathered at the MAB meeting in Boston to help prepare for the Third International Research Symposium.

Baumbach will serve on the committee. Dr. Shy is in the process of recruiting other members for the committee. Chris Pentz of Pentz Healthcare Conference Management was an invited guest at the MAB meeting. Chris has been retained by the CMTA to plan the logistics for the meeting.

Each member of the CMTA's Medical Advisory Board gives his or her time as a volunteer to review grants, plan seminars, and answer patient questions for the newsletter while maintaining an active clinical or research life. The CMTA is grateful for their generous volunteer contributions.

podiatrist is needed; however, this brace can surely fit the need for many people.

This brace is definitely for support and stability for people with drop foot and lower leg weakness secondary to CMT. Owing to the relatively low cost and ease of wearing, it is something that should be considered when choosing bracing or supports for an individual with CMT.

The brace is available through a physician or it can be obtained directly from Bauerfeind USA, Inc., 2100 Barrett Park Drive, Suite 508, Kennesaw, GA 30144. 1-800-423-3405.



CMTA MEDICAL ADVISORY BOARD

Shukuro Araki, MD
Kumamoto Medical School,
Kumamoto, Japan

Lisa Baumbach, PhD
University of Miami,
Miami, FL

Jose Berciano, MD
Universidad de Cantabria,
Spain

Thomas Bird, MD
VA Medical Center,
Seattle, WA

Pierre Bouche, MD
Neurologic Hospital
of the Salpêtrière,
Paris, France

Walter Bradley, MD, FRCP
University of Miami,
Miami, FL

Mark Bromberg, MD
University of Utah,
Salt Lake City, UT

Phillip Chance, MD
Children's Hospital
of Philadelphia
Philadelphia, PA

**J.S. Chopra, DCH,
FRCP, PhD, FAMS**
Post Graduate Institute
of Medical Education
and Research,
Chandigarh, India

Michael Conneally, MD
Indiana University,
Indianapolis, IN

Peter Dyck, MD
Mayo Clinic,
Rochester, MN

Francis Dyro, MD, FAAN
Portland, ME

King Engel, MD
USC,
Los Angeles, CA

Kenneth Fischbeck, MD
University of Pennsylvania,
Philadelphia, PA

Carlos Garcia, MD
Louisiana State University
Medical Center,
New Orleans, LA

Hans Goebel, PhD
University of Mainz,
Mainz, Germany

John Griffin, MD
Johns Hopkins University,
Baltimore, MD

Ludwig Gutmann, MD
University of West Virginia,
Morgantown, WV

Linton Hopkins, MD
Emory University,
Atlanta, GA

John Hsu, MD
University of
Southern California,
Downey, CA

Victor Ionasescu, MD
University of Iowa,
Iowa City, IA

Jan Korthals, MD, PhD
University of South Florida,
Tampa, FL

Roger Lebo, PhD
Boston University
School of Medicine
Boston, MA

Robert Lovelace, MD, FRCP
Columbia University,
New York, NY

James Lupski, MD, PhD
Baylor College of Medicine,
Houston, TX

**Kieran T. Mahan,
DPM, FACFS**
PA College Podiatric
Medicine,
Philadelphia, PA

**Harold Marks, MD,
FAAN, FAAP**
Al duPont Institute,
Wilmington, DE

Richard Mayer, MD
University of Maryland,
Baltimore, MD

James Mcleod, MD
University of Sydney,
Sydney, Australia

Jerry Mendell, MD
Ohio State University
Columbus, OH

Lefkos Middleton, MD
The Cyprus Institute of
Neurology & Genetics,
Nicosia, Cyprus

Robert Miller, MD
UCSF,
San Francisco, CA

Stanley Meyers, MD
Columbia University,
New York, NY

Carol Oatis, PT, PhD
Beaver College
Glenside, PA

Shin Oh, MD
University of Alabama,
Birmingham, AL

Gareth Parry, MD, FRACP
University of Minnesota,
Minneapolis, MN

Jack Petajan, MD, PhD
University of Utah,
Salt Lake City, UT

**Irena Hausmanowa
Petrusewicz, MD, PhD**
Warsaw Medical Academy,
Warsaw, Poland

Roger Rosenberg, MD
University of Texas,
Dallas, TX

Marvin Rozear, MD
Duke University,
Durham, NC

George Serratrice, MD
University of Marseille,
Marseille, France

Ding Guo Shen, MD
Chinese PLA
General Hospital,
Beijing, China

Michael Shy, MD
Harper Hospital, Wayne
State University,
Detroit, MI

Betty Soliven, MD
University of Chicago
Hospital
Chicago, IL

Ulrich Suter, MD
Institute of Cell Biology,
Zurich, Switzerland

Thomas Swift, MD
Medical College
of Georgia,
Augusta, GA

P.K. Thomas, MD, DSc
Royal Free Hospital,
London, England

Jeffrey M. Vance, MD, PhD
Duke University
Medical Center,
Durham, NC

Gerald Weber, DPM
Columbia University,
New York, NY

Kirk C. Wilhelmsen, MD, PhD
UCSF,
San Francisco, CA

Lowell Williams, MD,
Retired

Hitoshi Yasuda, MD
Shiga University of
Medical Science,
Shiga, Japan

Cycling for CMT

By Joe Beernink

Long before I became a member of the CMTA's Board of Directors in 1996, I realized that CMT was as much of an emotional and psychological challenge as it was a physical one. When I was diagnosed with CMT, I was told that there was a great chance that I would be in a wheelchair when I was 20, and that I should not plan to start any hobbies requiring physical exertion. That was 19 years ago. This year, at age 25, I am training to ride in the Rocky Mountain Cycling Club's (RMCC) Denver-to-Aspen Classic.

The Denver-to-Aspen Classic is a 1-day, 200-mile ride through the Central Rockies, crossing 10,001-foot Kenosha Pass, 10,051-foot Red Hill Pass, 9,346-foot Trout Creek Pass, and the monster 12,095-foot Independence Pass. We depart from Denver at 3:30 AM on July 26th and must be in Aspen by 9:30 PM the same day. It is a punishing and demanding bicycle ride that is as much a test of will power as physical endurance.



Joe Beernink trains for the rigorous Denver to Aspen Classic which he will use to raise money for CMT research.

Use this form to make a pledge toward Joe Beernink's ride in the Aspen Classic. All pledges will go toward CMT research.



With the support of the RMCC and the CMTA, I have chosen to use my participation in this event to raise money for research into CMT. Every dollar I raise in this effort will go toward summer research grants awarded by the CMTA to qualified applicants for 1998, in the hopes that their work will bring us all closer to a cure.

But the fund raising is not the only aspect of this ride. I receive many letters every week from people with CMT and their families who wonder how to cope and wonder what is ahead for themselves and for their loved ones with the disease. I want this ride to demonstrate that life does not end with CMT, and that with hard work someone with CMT can do things that many "healthy" people would never attempt.

So, I ask you to spread the word about my ride and to recruit sponsors. You can sponsor me by the mile, by my average speed while on the bike, by my top speed, or whatever. Or, just give a flat amount. There is no guarantee that I will be able to finish the ride. Many people don't. But plenty do, and I have been training aggressively for it, and should be ready by the time July 26th rolls around.

Even if you can't give any money, spread the word that I am doing this. Let everyone know that the strength of the human spirit can overcome the highest peaks and the steepest climbs. And that CMT *can* be beaten, as long as we keep the rubber side down and keep cranking.

For more information on the Denver-to-Aspen Classic, or the RMCC, visit their web page at <http://www.hemi.com/~rmcc> or write to them at Rocky Mountain Cycling Club, PO Box 101473, Denver, CO 80250-1473.



Cycle for CMT Research



PLEDGE FORM

The names on the form listed below agree to sponsor Joe Beernink in the Rocky Mountain Cycling Club's 1997 Denver-to-Aspen Classic, a 200-mile bicycle race to take place on July 26, 1997. All money donated to sponsor Joe in this event will go toward the CMTA's 1998 Annual Summer Research Grants for Graduate-Level Research into the treatment of CMT.

It is understood that it is not necessary for Joe Beernink to complete the entire ride, but a minimum of 1/2 of the ride (100 miles) must be completed within the 18 hours. Should the event be postponed to a different date within the 1997 calendar year, the pledges will remain valid. Should the event be canceled or should Joe not participate in the event, all pledges will be void.

Donors will be notified of the results of the ride by mail within 4 weeks of the completion of the ride. Questions should be directed to the CMTA at 1-800-606-CMTA. All pledge forms should be returned to the following address: Cycle for CMT 1, c/o The Charcot-Marie-Tooth Association, 601 Upland Ave., Upland, PA 19015-2494.

In order to provide a realistic expectation, the following maximum values have been set: Maximum miles are 210. Maximum hours on the bike are 20. Maximum average speed is 25 mph.

Sponsor Contact Information	Flat Amount Pledged	Rate Per Mile	Rate Per MPH	Rate Per Hour
Name _____ Address _____ Phone _____				
Name _____ Address _____ Phone _____				
Name _____ Address _____ Phone _____				
Name _____ Address _____ Phone _____				
Name _____ Address _____ Phone _____				

Disclaimer: No money will be collected in advance of the ride. The approval for Joe Beernink to use the Denver-to-Aspen Classic as a charitable event has been granted by the Rocky Mountain Cycling Club. This does not mean or require that any other riders will be riding for the CMTA, nor does it grant the CMTA the exclusive right to use the Denver-to-Aspen Classic as a charitable event. There is no affiliation between the Denver-to-Aspen Corporate Sponsors and the CMTA or Joe Beernink.

Chris Pentz and Paul Flynn attended the Medical Advisory Board meeting of the CMTA, which was held in conjunction with the American Academy of Neurology conference. Board members discussed plans for the Third International Research Symposium on CMT Disorders.



Research Symposium Will Be A Landmark Event

Preparation has begun in earnest for the Third International Research Symposium on Charcot-Marie-Tooth Disorders, which is to be held on October 22-25, 1998, in Montreal, Canada. Given the scope and importance of this project, the CMTA Board of Directors determined that professional expertise would be required to facilitate this undertaking. In April, the Board engaged the services of Ms. Chris Pentz of Levittown, PA and Mr. Paul Flynn of Newark, NJ.

A certified meetings professional, Chris Pentz has acquired extensive experience during her 15 years in the corporate world, working for several Fortune 500 Companies. Chris started her own company about 3 years ago, specializing in creating, planning, and managing professional programs designed to support corporate efforts to educate and inform specific audiences on a message or product, using press, group communication forums, and exhibits as vehicles.

Chris is a long-time member of Meeting Professionals International (MPI), and she is currently a member of the International Marketing Committee. Chris is also very active at the chapter level, where her peers voted her Philadelphia Area Chapter Planner of the Year in 1994, and Chapter Manager of the

Year in 1995. She is currently the President of the Philadelphia Area Chapter of MPI.

Prior to assuming the role of Development Coordinator, which necessitated his resignation from the Board, Paul Flynn served the CMTA as chairman of the fundraising committee and as a member of the marketing and public relations committee. His primary responsibility is to secure sponsorship funding for the research symposium while assisting the Board with other fundraising programs, such as those for research and operating expenses. This is a part-time commitment that he balances with his job as Acting Director of Development at Seton Hall Law School. Corporations or individuals interested in funding the symposium should contact Paul Flynn through the CMTA office.

Joe Beernink, a new Board member from Colorado, employed by Neodata, a subsidiary of EDS, is serving as the chairman of the ad-hoc committee for the research symposium. In addition to coordinating the activities of the committee, Joe will work closely with symposium chairman, Dr. Michael Shy, a Medical Advisory Board member from Wayne State University in Detroit, MI. In his preliminary outreach for presenters, Dr. Shy received very positive responses from his colleagues. "I am extremely pleased by the interest shown for this symposium," Shy said. "Researchers from around the world are very excited about this gathering...this will be a landmark event."

CALL FOR PARTICIPANTS:

The laboratory of Dr. Lisa Baumbach at the University of Miami has recently discovered a new DNA polymorphism in the pmp-22 gene in the CMT1A duplication region. The polymorphism appears to be specific to individuals of African-American heritage and was found in conjunction with other disease-causing mutations in two unrelated severely affected CMT patients of African-American descent. Dr. Baumbach's laboratory is requesting blood specimens from any CMT or Dejerine-Sottas patients of African-American heritage for further molecular studies related to this new polymorphism and genotype:phenotype correlations. For further information, please contact Dr. Lisa Baumbach, University of Miami School of Medicine, RM 6021 MCCC, 1601 NW 12th Avenue, Miami, Florida 33136; 305-243-3997 (phone); 305-243-3919 (fax); E-mail: lbaumbac@mednet.med.miami.edu.

Shriners Hospital for Children in St. Louis Hosts Family Conference on CMT

On April 5, 1997, the CMTA, in conjunction with the Shriners Hospital for Children in St. Louis, Missouri, held the first 1997 family conference on CMT. Approximately 75 people were in attendance and experienced the small group session format for the first time. Participants heard a presentation by Dr. Michael Shy of Wayne State University in Detroit on the most recent genetic breakthroughs and were encouraged by his information that the peripheral nervous system can regrow nerves and is, therefore, much better than the central nervous system at repairing itself. Next, the audience heard Vincent Bertolino of Athena Diagnostics discuss when one might consider having the diagnostic DNA test for type 1A or type 1X. After these talks, the attendees could choose to attend small group sessions on physical therapy and occupational therapy, pain and the neurotoxic drugs, or parenting concerns for families with CMT.

Following lunch, Dr. Perry Shoenecker, Chief of Staff and an orthopaedic surgeon at Shriners, discussed surgical options for children with CMT and Dr. Jeffrey Johnson of Washington University discussed surgeries for the adult population. The group then moved into the small group sessions, choosing a different

topic area from what they had in the morning.

The smaller groups allowed more questions and answers to be handled and provided opportunities for the speakers to use visual aids and demonstrate exercises.

The small group sessions were rated very highly on the survey sheets completed after the conference and plans are underway to continue that format at the next conference in October, 1997 in Detroit.

On Friday night, April 4, 1997, approximately 40 persons attended the hospitality suite at the Frontenac Hilton, which was hosted by Athena Diagnostics and the members of the Kansas City, Missouri, support group. The get-together provided an excellent opportunity for people to get to know one another and to ask questions of the doctors and therapists from Shriners who attended.

The conference plans and the hospitality suite were arranged by Ardith Fetterolf, CMTA Board Member and Kansas City support group leader. Dr. Perry Shoenecker and Becky Hudson handled the arrangements for the facilities and the speakers for the conference. The CMTA thanks these volunteers for their help in making the conference such a success.

Orthotist Eddy Fuentes conducts a small workshop on shoe inserts and bracing during the St. Louis conference.





**YOU CAN REACH
CMTnet
ON THE INTERNET**

**@URL<[http://www.
ultranet.com/
~smith/
CMTnet.html](http://www.ultranet.com/~smith/CMTnet.html)>**

Shriners Hospitals for Children: "World's Greatest Philanthropy"

Shriners Hospitals, located in the United States, Canada, and Mexico, are operated by the Shrine of North America. There is never a charge to patients for any of the medical care or services received at Shriners Hospitals, including outpatient and inpatient care, diagnostic services, surgery, medical care, casts, braces, X-rays, rehabilitation programs, recreation, and continuation of their education.

Children from infancy to their 18th birthday may be eligible for treatment if, in the opinion of the chief of staff, there is a reasonable possibility that treatment will benefit the child and if treatment at another facility would place a financial burden on the patient's family or guardian. Shriners Hospitals for Children are open to children regardless of their race, religion, or relationship to a Shriner.

In 1922, when the first Shriners Hospital opened in Shreveport, LA, it marked a commitment the Shrine of North America has continued for generations—helping children build better lives. The Shrine is an international fraternity that has built 22 Shriners Hospitals, 19 for orthopaedic care and three burn centers. In addition to offering a complete range of specialized medical and rehabilitative services, Shriners Hospitals use a unique family-centered treatment approach that involves the patient's family in the recovery process. This philosophy stresses that while medicine might heal the child's body, tending to the child's sense of well-being is equally important to recovery.

Shriners Hospitals are equipped and staffed to care for children with congenital orthopaedic deformities and problems resulting from diseases of the bones, joints, and muscles. Some of the common orthopaedic problems treated at Shriners include:

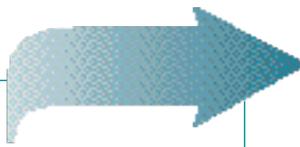
- Scoliosis (curvature of the spine)
- Hand problems
- Back problems
- Hip dysplasia
- Orthopaedic problems resulting from neuromuscular disorders

Each year, Shriners Hospitals spend millions of dollars on specialized research and the development of new treatments for crippling diseases that affect children. Shriners Hospitals are highly

regarded as leaders in the field of orthopaedic medicine, surgery, and research, and a great many orthopaedic surgeons receive their training at Shriners' facilities.

Application forms can be obtained from any Shriner, Shrine Temple, Shriners Hospital or by writing to Shriners Hospitals for Children, P.O. Box 31356, Tampa, FL 33631-3356. For general admission information, call 1-800-237-5055 (in Canada call 1-800-361-7256).

Shriners Hospitals are located in Chicago, IL; Erie and Philadelphia, PA; Greenville, SC; Honolulu, HI; Houston, TX; Intermountain, UT; Lexington, KY; Los Angeles and San Francisco, CA; Mexico City, Mexico; Portland, OR; St. Louis, MO; Shreveport, LA; Spokane, WA; Springfield, MA; Tampa, FL; Twin Cities, MN; and Montreal, Quebec, Canada.



SAMPLE LETTER FOR INCLUSION IN STUDENT FILES

One of the excellent handouts from the conference at the Shriners Hospital in St. Louis, Missouri, was the following letter created by Loray Dailey, Director of the Occupational Therapy Department, Cindy Steiner, Director of Physical Therapy, and Emery Miko, Director of Prosthetics and Orthotics.

SAMPLE LETTER FOR INCLUSION IN STUDENT FILES

Dear Teacher,

(Child's Name) has Charcot-Marie-Tooth Disorder. This is a familial progressive motor and sensory disorder which affects the peripheral nerves. It does not affect life span, the heart or intelligence, but there is no cure. Research shows the nerves lose their covering and this causes the nerve impulses to the muscles to slow down. The nerves to the feet and lower legs and the hands and the forearms are the only ones affected in this condition.

Charcot-Marie-Tooth starts in childhood or adolescence. It may first be recognized because of various symptoms such as foot deformities, clumsy gait, difficulty running, developmental delays, tremors in the hands or feet and, later, loss of finger dexterity. It affects the feet and lower legs first, and then the hands and forearms. There is muscle weakness, muscle imbalance, joint deformities and loss of sensation for pin prick and temperature and proprioception in the feet and hands in a "stocking and glove" distribution. Other orthopaedic problems may include scoliosis and hip abnormalities. This leads to loss of function for patients.

In extreme forms of CMT, a child may lose his/her ability to walk or use his/her hands normally. Treatment is symptomatic and includes bracing, therapy, surgery and educational and psychological support.

The services of the physical and occupational therapist are essential at school to manage and optimize function. These goals are medically essential to manage Charcot-Marie-Tooth in the school setting:

1. Maximize Strength

- Daily exercise program
- Adapted physical education
- Aerobic/cardiovascular program such as riding a stationary bicycle or swimming
- Studies have demonstrated improved function in patients with neuropathies following a prolonged exercise program.

2. Ensure Safety

- Environmental/functional evaluation of school environment

For example: Ankle weakness may cause walking on an irregular surface to be dangerous in terms of falling with secondary risks of sprains or fractures. Identifying the need for an elastic ankle support may prevent injury at school. Grab bars at different heights in the bathroom may be needed to prevent falls due to poor balance.

3. Protect Joints

- Writing with a fatter pencil (or a pen instead of a pencil) to decrease muscle cramps and hand pain.
- Thumb splints to provide support or stabilization to improve grasp strength for functional activities at school.

4. Energy Conservation

- Use a tape recorder for homework assignments.
- Modify class schedules to reduce distances between classrooms.
- Allow rest periods at school or on field trips.
- Provide keys for elevators.
- Use motorized scooter.

5. Family Support

- Parents know their child...work cooperatively with them to problem solve.

Thank you for your interest, help and support.

Sincerely,

Physician

Therapist

This letter is included here as an example of what parents might want to solicit from their child's physician or therapist to be included in their student's file at school. It answers many questions regarding CMT and what problems a child might experience, and it offers concrete examples of how a school can help accommodate a child with CMT.

UPCOMING CONFERENCE ALERT:

A patient/family conference is scheduled for Detroit, Michigan, on October 17 and 18, 1997. The host of the conference will be Dr. Michael Shy of Wayne State University School of Medicine, a member of the CMTA Medical Advisory Board and Program Chair of the Third International Research Symposium on Charcot-Marie-Tooth Disorders. There will be a hospitality suite on Friday night in the atrium of the research building at Wayne State and the conference will take place on Saturday. In addition to conventional presentations, there will be five workshops, one of which will be a tour of the research facilities at Wayne State, where Dr. Agnes Jani is currently using a CMTA post-doctoral grant of \$35,000 to continue working on gene research.

Two other conferences are tentatively planned for 1998. They are in the planning stages for Miami, Florida, under the auspices of Dr. Lisa Baumbach and Dr. Walter Bradley, and in Seattle, Washington, hosted by Dr. Thomas Bird.

Why Should You Have a CMT DNA Test?

By Vincent Bertolino, Athena Diagnostics

Clinical diagnoses are statements of probability.¹ If your nerve conduction velocity is slow; if you have had problems with your feet for years, and if your hands are now starting to become weak, you probably have CMT1. Furthermore, since CMT1A is the most common variant, you are most likely to have type IA. Probabilities and likelihoods are what clinical diagnosis is about, in the absence of definitive diagnostic testing.

Probabilities are fine for describing populations or developing theories about future events, but what good are statistics to an individual diagnosis? How would you feel if your physician said that there was a good chance that your sore throat was caused by an infection, that there was a 50% chance it was strep throat, but then did not offer you a test to find out? How would the result affect your care? How would your care be affected by the absence of a test?

The strep throat example is fairly trivial since therapy and prognosis are very different in each case of strep, cold, or allergy. There is an obvious need to test when you have a sore throat that could be caused by a strep infection. Therapy exists; the disease is contagious and dangerous;

therefore, it must be identified and promptly controlled.

Does the need to perform a diagnostic test change if the disorder in question has no cure or if patient management by the physician is not likely to be changed as a consequence of the result of the test? The answer depends on the circumstances of each case, but in general, as in the sore throat example, *more information is better patient management.*

How having more information about your CMT variant will affect your life depends on you and upon your circumstances. Are you satisfied with your clinical diagnosis? What would you do differently if you knew your variant with certainty? How would you feel after acquiring the new information? Will the knowledge be helpful for your parents or your children? How would it affect your insurability or employability?

One motivator for being tested is personal preference. You will have your genes for life and if you want to know all you can about them, that is your choice. This is very much an issue of your personal quality of life or peace of mind.

In the case of a child suspected of having CMT, it may be preferable to have a blood test

CMTA Awards Four Summer Research Grants

The Charcot-Marie-Tooth Association in conjunction with its Medical Advisory Board announced in April that four fellowships had been awarded in a variety of fields for CMT research. Each award is a \$4,000 grant to enable a student to work during the summer in the laboratory of an established researcher or clinician.

The first recipient of a summer grant was Mr. Spencer Blackman who will enter the University of Miami Medical School in the fall of 1997 and is working with Dr. Lisa Baumbach on "Genetic Anticipation in Charcot-Marie-Tooth Disease: Continued Molecular Investigation of the CMT1A Locus." His work will continue the work begun in 1995 with a similar grant from the CMTA. The research at Miami continues to focus on the

hypothesis that an increasing severity of disease presentations within kindreds is associated with DNA rearrangements that disrupt a number of normal genes in the CMT1A duplication region on chromosome 17p11.2, thus giving rise to different disease manifestations.

The second award winner is Dr. Eleni Zamba of the Cyprus Institute of Neurology and Genetics. Dr. Zamba will be working at the laboratory of clinical neurophysiology at the Neurological Institute in New York City. She will continue the work of the Cyprus Institute focusing on collecting families with CMT2 and recessive CMT for evaluation aimed at identifying additional genes and proteins involved in the pathogenesis of CMT.

instead of the electrodiagnostics (EMG/NCV) traditionally used to make a diagnosis. Drawing a blood specimen is less painful and fairly quick; it also has the advantage of being highly specific and sensitive.

Denial of insurance coverage on the basis of one's genetic tests has been a significant concern. This is a complex issue beyond the scope of this article, but there are several practical points you should consider while consulting with your family and your medical advisors. If your medical chart contains a diagnosis and history of CMT, then you already have genetic information in your life regardless of whether you also get tested. This is true any time a physician takes patient and family history for any disorder (cholesterol, heart disease, cancer, etc.) For this reason, care should be exercised in applying DNA tests to family members who may be carriers, but who have not developed clinical signs of CMT.

It is conceivable that a DNA test could help a family member get insurance coverage. For example, if a family has been tested and found to be positive for CMT1A, it would be very unlikely that any other CMT gene would be involved in the affected family members. If an asymptomatic member of this family were to be denied coverage based on a family history of CMT1A (pre-existing), a negative DNA test could be proof that this patient was not subject to the pre-existing family history of CMT1A.

The third recipient is Odile Dubourg, who is a consultant in clinical neurophysiology at the Hospital de la Salpetriere in Paris, France. He works with Dr. Pierre Bouche and will be studying a large series of 210 CMT pedigrees collected at La Salpetriere Hospital. He plans to classify the families according to patients' motor nerve conduction velocities and mode of inheritance and to evaluate frequencies of 17p11.2 duplication and PMP22, P₀ and Cx32 mutations in those subgroups. The data will permit the elaboration of a rational strategy for molecular diagnosis based on median MNCV (median nerve conduction velocity) and mode of inheritance.

The final recipient is Jeffrey Bonneville, a certified orthotist and a medical student at the University of Health Sciences, St. Johns, Antigua. Mr. Bonneville plans to produce a usable standard for the application of orthoses to the CMT population, and by disseminating this information to the medical community, raise the awareness of CMT as a diagnostic

Knowing your variant of CMT with certainty will tell you how the trait could be inherited, and under what circumstances it would not be passed on. For instance, if you are a male and your test result is positive for CMT1X, and your wife does not have CMT, you can expect that none of your sons will have CMT1X, and that all of your daughters will be carriers of CMT1X.² Under these specific circumstances, your children would not need to be tested.

Another benefit of knowing which specific gene you have is to be able to follow the research. Different investigators focus on different types of CMT. As new information is published, you can know what is most applicable to you, which might include clinical trials or new therapies.

Regardless of what variant of CMT you may have, the quality of your life is in your hands. You decide to make the effort and seek the knowledge and training needed to work at keeping as much function and strength as possible. You decide to communicate with others who have CMT and find out what works for them. You decide to become a member of the CMTA, follow the research, and become more knowledgeable about CMT.

Should you get tested? You decide.

¹ Saunders, A.M., et.al. Lancet 1996;348:90-93.

² Parry, Gareth, J. Charcot-Marie-Tooth Disorders: A Handbook for Primary Care Physicians. 1995. Charcot-Marie-Tooth Association, Upland, PA.



Vincent Bertolino of Athena Diagnostics spoke at the St. Louis Conference about the diagnostic DNA test for CMT1A and IX available from his company.

group. He plans to develop a national questionnaire that will be submitted to 2,500 members of the American Board of Certified Orthotics and Prosthetics and the American Academy of Podiatry via professional publications, the Internet, fax, and mail. The questionnaire will help correlate the types of orthotic devices, the age and activity level of CMT patients, and the specific use of certain devices. Mr. Bonneville has been an orthotist in private practice for 19 years.

In an effort to solicit proposals from a wider variety of specialties, CMTA Board Member Louise Smith, chair of the research committee, sent letters announcing the grants to schools offering graduate work in physical therapy, orthotics, and prosthetics, as well as to all the members of the Medical Advisory Board of the CMTA. The variety of topics for the grants as well as the locations from which they came indicates that the scope of the CMTA's research efforts is continually increasing.

Contributions to Our 1997 Research Appeal

The CMTA thanks these donors for their support. The 1997 Research Appeal marks the second year of the Armington Challenge, during which time the CMTA hopes to improve on the success of the first year of the challenge. If you have not yet contributed, please consider a gift to research...it's the hope of the future. Additional gifts will be listed in the summer newsletter.

\$5,000 AND OVER GIFTS

Stanley and
Phyllis Sanders

\$300-\$4999 GIFTS

Anonymous
John Chernega
Ron L. Davis
Diane M. Freaney
Gary A. Kauffman
J. Rodman Steele, Jr.
Surgimed - MLB, Inc.
Nancy D. Touhey

\$100-\$299 GIFTS

Susan B. Adams
Kirsten K. Bergin
John K. Best
Carol Ann Bomberger
R. Gordon Bradwick
Joseph Burke
Brenda A. Callahan
Mark E. Choules
William F. Derr
Ford F. Farabow
William R. Foster
Dino W. Giamatti
Walter Goertz
Roger D. Grieser
Esther B. Haber
Helen B. Hardison

Jerry G. Idleman
Woody Kloesel
George Kunath
Robert D. Larsen
Michael E. Leeson
Sheila Levine
Ronald A. Lynch
Jean Ann Marshall
H. P. Maruska
Daniel Max
Marjorie McCallum
John C. Merion
Anthony V. Napoli
Angela R. Neal
J. Kendrick Noble
Jane Orlansky
Henry A. Silver
Robert L. Switzer
Robert G. Trumble
Jane S. Twaddell
Kathleen Weinstein
A. Hart Wurzburg

ALL OTHER GIFTS

Patricia J. Albury
Richard L. Andeel
Gunnard E. Anderson
Sue Atkins
John Bachkosky
Edward F. Bachman
Cheri N. Ball
Judith Beavers
Marsha Bennetto
Carol Benusches
Harry Berkovitz
Samuel Berry
Jack Beyers
Jill L. Boyer
Charlotte Brieff
Irvin M. Buckman

Lisa Burda
Robert Burt
Dominick M. Calsolaro
Justin M. Carisio
Peter L. Carter
Catherine M. Chinni
Kim Clark
Martha L. Clark
Earl F. Clay
Robyn L. Cohen
Denny Collins
Joe A. Collins
Zelda Connelley
Edgar Denton
Elsie Di Liberto
Tina M. Doyle
Nancy K. Dreher
Anne A. Dunn
Ruby B. Egnew
Margaret Elmer
Robert A. Ellsworth
Efrain Espada
Frances P. Festa
Lucille FitzPatrick
Charles A. Ford
Jean A. Foreman
John R. Forster
Anna G. Forte
Rosemarie R. Foss
Dan Fox
Helen Fox
Russell F. Frazier
Alice E. Freeman
Henry Friedmann
Andrew J. Geary
Joshua Gerbert
Carl E. Gluck
Phyllis Goodman
Michael L. Grant
Jack A. Graves
Frank Gussey
Robert C. Hailer
Ann E. Hall
Elizabeth A. Harbold
Earl Hays

Richard F. Hathaway
Jeanell M. Haynie
Roy A. Hayward
M. Delores Hedrick
Sandi Herschel
Donna D. Hess
David D. Hockett
Leo T. Hogan
Leah Holden
Herbert J. Holly
Bryn Housh
Theresa Hyde
Joan C. Iwasko
Sharon H. Jackson
Craig Jewart
Candace L. Johnson
Dennis L. Judd
C. Robert Karl
John R. Kathrein
Janis M. Kelly
Doris Kimpton
Anna Marie Klein
Robert Koch
Marie M. Kroninger
Joseph S. Kutzler
Steve J. Landgraf
Bert Layne
Daniel J. Leahy
Juanita K. Legg
Julie Leonard
James G. Leow
Leonard Lietz
Joel S. Lipset
Carol Sue Liston
Timothy J. Lloyd
Doris S. Lochner
Lois L. Lott
Dirk R. Lockett
Carol C. Madden
Deborah Maier
Pascal M. Malassigne
Connie A. Manson
H. Louise Marcus
Lawrence J. Mauceri
Donald E. McClean

Terry A. McIntosh
 Katherine McMahon
 Judi G. Michel
 Gerard E. Milne
 Kenneth Munger
 James F. Murdock
 Debra K. Nelsen
 Claire M. Nusi
 Madonna O'Dell
 Le Roy Oldack
 Ellen J. O'Neill
 Charmaine Orłowski
 Dennis L. Parker
 Frank Polenik
 Delta D. Raley
 Janice Rensing
 Sylvia Rider
 Raedel Rind
 Lewis R. Roberts
 David F. Rock
 Betty J. Roemig
 Richard J. Rogers
 Verna Rogers
 Lawrence Roman
 Lesia L. Rouse
 Norma R. Routledge
 Mary E. Ruotolo
 Barbara Saljian
 Stephen Sand
 Harry P. Schaller
 Victor E. Schaller
 Gregory G. Scham
 James D. Schoonover
 Peter Schroeck
 Janice Schulman
 Stephen B. Scofield
 Alex Segal
 Carol M. Shatto
 Judi Shields

Ruth Silver
 Robert L. Smallwood
 Peter L. Speronis
 Bette F. Staub
 Ruth E. Stefanovich
 Susan R. Steingarten
 Mark V. Stephan
 Doris V. Stroschin
 Willaim D. Sullivan
 Rosemary C. Talarico
 K. Laurie Terry
 William H. Thompson
 Richard J. Tichenor
 Homer I. Titus
 James W. Tucker
 Louis K. Tullock
 Janice A. Turek
 Harlan E. Van Engen
 Arnold N. Wagner
 Lester L. Ward
 Holly K. Waters
 Clyde C. Weger
 Rita M. Welsh
 Michael A. West
 Betty Jean Wilkin
 Geneva C. Williams
 Lowell L. Williams
 Marjorie L. Williams
 Susan A. Williams
 Robert A. Williamson
 Harold M. Wilson
 Patty Wilson
 Andrew Winokur
 Randolph W. Wise
 Beverly B. Yanke
 Mary Elizabeth York



The CMTA Opens a Home Page!

If you are one of the many fans of the World Wide Web, you can now find information about the Charcot-Marie-Tooth Association on its own home page at www.charcot-marie-tooth.org. The site is still under construction, but you can find profiles of some of the members of the Board of Directors and the employees of the organization. Until now, we have relied on CMTnet, the home page developed and maintained by CMTA Director Louise Smith, as the vehicle to enable people to find out about our organization and our publications. We will still remain linked to her address and she to ours because the two sites vary tremendously in content. Louise has always offered valuable research information and continues to do so. The CMTA's home page is more personal and is intended to introduce someone new to the organization to what we do and who we are. Sample articles from our newsletters, past and present, are included to provide insight into the style and content of our publications.

We continue to offer answers to questions through our new E-mail address at CMTA@charcot-marie-tooth.org and we encourage our members to send in medical questions they would like to see answered in the newsletter. All medical questions are sent to members of our Medical Advisory Board for qualified answers.

If you have suggestions for things you'd like to see included on our home page, please send your thoughts to us by E-mail and we will respond promptly.

While a great deal of effort has been made to make this list as accurate as possible, we apologize for any omissions or misspellings that may have occurred.



WRITE TO US!

Pat Dreibelbis, Editor

The CMTA Report

CMTA

601 Upland Ave.

Upland, PA 19015

The CMTA Report

is published by the Charcot-Marie-Tooth Association, a registered non-profit 501(C)(3) health organization. Copyright 1997, The CMTA. All rights reserved under International and Pan American Copyright conventions. No part of this newsletter may be reproduced in any form or by any electronic or mechanical means, including information storage and retrieval systems, without permission in writing from the publisher.

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.

Ask the Doctor

Dear Doctor,

Is CMT associated with vocal cord paralysis? If one cord has been determined to be paralyzed, is the other certain to follow?

The Doctor replies:

Dr. Peter Dyck of the Mayo Clinic in Rochester, MN, has published a report on a family with CMT type II in which there was vocal cord paralysis associated with their CMT. However, there have been no cases of vocal cord paralysis in type I. The first question, then, would be what type of CMT do you have? In the family (from Arkansas) with the vocal cord paralysis, there was very early onset of the disorder, paralysis of the diaphragm as well as the vocal cord, and the second vocal cord was also paralyzed.

You should be seen by an ear, nose and throat specialist. He or she will make the diagnosis by the process of exclusion, that is, eliminating all other possibilities and connecting the paralysis to your CMT only if everything else can be ruled out.

If you would like further information, you could contact the Mayo Clinic or write to the CMTA with more details about your CMT (type) and your vocal cord problems and a member of the Medical Advisory Board will try to help you.

Dear Doctor,

About 5 years ago, after 45 years of smoking, I quit. For several years prior to cessation of smoking and from that time forward, I have had a productive cough. I visited a pulmonary specialist who did all the capacity tests and concluded with an endoscopic tour of both lungs, all lobes. His conclusion was, vital

Letters to the Editor:

Dear CMTA,

My husband has CMT and we agreed early in our relationship that we would not conceive children for fear of passing on the CMT. We both wanted to have a family and tried unsuccessfully to adopt for several years. During that time, I heard about a process called pre-implantation genetic testing that was being done at Baylor in Houston, Texas. Through the process of in vitro fertilization, a woman's eggs were removed and fertilized with the husband's sperm outside the womb. Without destroying the fertilized egg, doctors could remove one cell from each embryo and test that cell for cystic fibrosis. The fertilized embryos unaffected by that disease were then returned to the mother's womb, where they would hopefully implant and mature. I immediately thought that if they were able to test for cystic fibrosis, perhaps they would be able to do the same for CMT.

We began to investigate the possibilities of going through this process ourselves, but at the same time, we did not know which type of CMT my husband had. The test for 1X became available on August 1, 1995, through Athena Labs, and I think we were probably first in line. The result was positive and we immediately contacted Baylor. Because there is no male-to-male transmission with type 1X, our goal was to conceive only boy children. Although Baylor could have

tested for the CMT itself, they explained to us that the signal is stronger for sex and thereby more easily and accurately determined.

In January 1996, I started taking the hormone injections that are part of the in vitro procedure. On February 13, 1996, I had a positive pregnancy test and in October, our beautiful son was born.

It is impossible to express how grateful we are to all of the people who work so diligently researching this disease to gain the knowledge to make things like this possible. We pray that the next step will be a cure and are thankful that there is now a means available to keep CMT from affecting our future generations.

—A Grateful New Mother

This letter is in reponse to D.B., whose letter appeared in the Summer/Fall issue of 1996.

Dear CMTA,

I was really excited to read your letter regarding magnesium and nocturnal leg cramps. My situation differs from yours slightly because I don't have leg cramps, but muscle "twitches," mostly in my legs but sometimes in my arms and hands as well, and was only taking magnesium as a regular but coincidental supplement.

I had discontinued taking the magnesium for a period of time, during which I noticed that these "twitches" were increasing in frequency and

capacity was slightly below normal, but not much, and lungs were clear and sound. Now, 3 years later, I still have what appears to be chronic bronchitis. My question is, if the cough and occasional shortness of breath may be caused by CMT, is there anything that can be done to reduce the coughing?

The Doctor Replies:

The coughing is probably not related to your CMT. It would appear that you do have bronchitis or some other inflammation that is probably a residual of smoking or some other irritant.

Dear Doctor,

My son, 4 years old, has been diagnosed with CMT type I. We have witnessed on two occasions, the most recent last night, that he suffers a dramatic, and I mean frightening, decline in function when he is suffering from a cold, especially when he has a fever. Although walking is always difficult, standing becomes impossible during these times. In fact, his

disability becomes so pronounced that he is unable to move his legs, even when lying down.

Last night, his temperature reached 102°F for a short period of time, during which he was unable to move his arms, too. I would like to know if others have this experience. I should add that after the first episode like this, it took almost 2 full weeks for him to recover his strength. But, he did recover. Is this normal in CMT?

A pediatric neurologist replies:

It is not unusual for children with neuromuscular disorders, including CMT, to have transient mild weakness associated with febrile illness, especially if the child is treated with bed rest. It would be very unusual, however, for a child with CMTI to have severe weakness lasting up to 2 weeks with a febrile illness.

I would suggest that you tell your pediatric neurologist about these episodes of transient severe weakness. He may want to do further tests to exclude disorders known to cause periodic paralysis.



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

intensity. They had gotten so bad that they would awaken me from a sound sleep with a start several times during the night. I then tried a mail-order homeopathic combination called Valerin, which advertised "natural relaxant for the relief of muscle spasms, tension, and stress. Valerin consists of valerian root, passionflower and magnesium carbonate. After only a couple of weeks, I noticed that I was sleeping through the night. My episodes of night "twitching" were again within the sleep toleration level.

Connecting the possibility that diminished magnesium might be the culprit, I once again started taking magnesium as a regular supplement (800 mg. chelated, spaced throughout the day) along with the two Valerin at night. Now my daytime "twitches" are also back to being almost totally contained.

I thought that this was all in my imagination because my neurologist couldn't see any connection between my "twitches" and the magnesium, so it is very reassuring to know that your experience is similar to mine. —M.D. Washington

Wonder Laboratories, the supplier of Valerin, can be reached at 1-800-992-1672. They have catalogs.



Dear CMTA,

I am an 18-year-old college student at Northeastern University and a month ago I went to my podiatrist complaining of pain in my great toe. It didn't look right even for my deformed feet. He told me that I had pulled the tendon in the top

of my foot. He gave me an equalizer at first, but my foot was too tiny and it slid around in the boot, therefore defeating the purpose. I then went to a cast, but the pain was so great that I had him take it off 2 days later.

The pain wasn't where the tendon was. It was under my foot where they put a metatarsal pad. When I was sitting still or sleeping, my foot rested there owing to the high arch and caused major pain. I went back to the equalizer for 4 weeks.

I went back to the doctor and he told me that not only was the toe worse than it had been, all of the other toes had gone "kaput" as well. That was the first major sign of CMT I have had besides the occasional aches in my feet and knees. He suggested AFOs and surgery at some later date because I am so young.

The problem is that I had already had AFOs for both my feet, but I had the same problem that I did with the cast. I had it readjusted, pads put in, had it sliced down, and nothing worked. The AFO lab must have been sick of seeing me. The AFO reduced the pain in my knees, but the bottom of my foot was unbearable and I couldn't take it anymore. I stopped wearing them.

My question is: has anyone had this problem and if so, how have they fixed it? Has anyone had surgery on the tendon and did it help more than hinder?

I would appreciate any suggestions because I am at my wit's end. —M.R.

Write to M.R. in care of the CMTA or E-mail her at Lrudge5@aol.com

The CMTA reserves the right to edit letters for space.

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.

What is CMT?

- ...is the most common inherited neuropathy, affecting approximately 125,000 Americans.
- ...is also known as peroneal muscular atrophy and hereditary motor sensory neuropathy.
- ...is slowly progressive, causing deterioration of peripheral nerves which control sensory information and muscle function of the foot/lower leg and hand/forearm.
- ...causes degeneration of peroneal muscles (located on the front of the leg below the knee).
- ...causes foot-drop walking gait, foot bone abnormalities, high arches and hammer toes, problems with balance, problems with hand function, occasional lower leg and forearm muscle cramping, loss of some normal reflexes, and scoliosis (curvature of the spine).
- ...does not affect life expectancy.
- ...has no effective treatment, although physical therapy, occupational therapy and moderate physical activity are beneficial.
- ...is sometimes surgically treated.
- ...is usually inherited in an autosomal dominant pattern.
- ...may become worse if certain neurotoxic drugs are taken.
- ...can vary greatly in severity, even within the same family.
- ...can, in rare instances, cause severe disability.
- ...is the focus of significant genetic research, bringing us closer to answering the CMT enigma.
- ...Type IA and CMTX can now be diagnosed by a blood test.

The CMTA Report

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



**Crozer Mills Enterprise Center
601 Upland Avenue
Upland, PA 19015
1-800-606-CMTA**

Forwarding and return postage guaranteed.
Address correction requested.

Non-Profit Org.
U.S. Postage Paid
Glen Mills, PA
Permit No. 10