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THE CMTA Report

Information on Charcot-Marie-Tooth Disorders for patients, families, and the scientific community ✲ www.charcot-marie-tooth.org

CMT and Ascorbic Acid

A Report from the Investigator funded by the CMTA

PROFESSOR MICHEL FONTÉS, Faculté de Médecine, Marseille, France

Our strategy was to develop animal models of genetic disorders in order to better understand human inherited disorders and to propose therapies. We developed, in collaboration with C. Huxley in London, an original approach to construct these models by inserting into the mouse genome the human region of our genome containing the gene involved in the pathology. The target of the medicine developed using the animal model is thus the same as human. Our first model concerned the Charcot-Marie-Tooth Type 1A disorder, and we used it to develop therapies. During a bibliography search, I observed that ascorbic acid (Vitamin C) was described as an absolute factor for myelination, so I decided to set up clinical trials on our CMT mouse. This approach allowed us to demonstrate that ascorbic acid at high doses stops the progression of the disorder and allows the recovery of muscle strength. Moreover, we understand how it works, allowing us to design new experiments in order to find second-generation drugs. Our results are thus opening the first human clinical trials on CMT patients. However, I would like to add the following lines in answer to questions that I have been asked by mail several times:

Regarding clinical trials of ascorbic acid in CMT1A, one must keep in mind that a molecule is not a medicine. We do not know if ascorbic acid works in humans and what the optimal dose is. We have an idea that ascorbic acid at low dose is inefficient. Mice differ from humans in that they synthesize ascorbic acid, and have, thus, a continuous level of circulating ascorbic acid, yet our CMT mice suffer from a neuropathy.

Moreover, we have indications that only very high doses are active. Thus, only the development of a true medicine will be efficient and this needs clinical trials. A protocol of a clinical trial has been written in France (Professor Olivier Blin is the coordinator). We already have regular approval of the trial through the competent governmental agency. We are, thus, trying to raise funds to do it. I hope that trials will start in France soon. We have also contact in USA to go on as quick as possible in this country.

At present, we have only tested CMTA mice and the

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THE CMTA REMEMBERS PRESIDENT ANN BEYER

Under Ann's leadership, the CMTA evolved into an important organization bringing together leading scientists and providing more information on CMT than any other organization in the world.

See page 4

INSIDE:

CMTA

Charcot-Marie-Tooth Association

OUR MISSION:

To generate the resources to find a cure, to create awareness, and to improve the quality of life for those affected by Charcot-Marie-Tooth.

OUR VISION:

A world without CMT.

EMAIL CMTA AT:
CMTAssoc@aol.com

Annual Research Appeal and New Research

CHARLES F. HAGINS, EXECUTIVE DIRECTOR

The Annual Research Appeal for the CMTA will be mailed in June, 2004 and it has an added significance that we hope to share with all donors.

On September 22-23, 2003, the CMTA Board of Directors agreed to adopt a New Research Strategy in conjunction with its Medical Advisory Board.

The objective of this New Research Strategy is as follows:

1. Broaden the scope of opportunities for research by extending relevant research beyond the $35,000 limit and/or the 12-month term.
Impact—Research studies often take place over the duration of several years and often we see applications for a second and third year from the same researcher. We believe that if the application is worth funding for one year it is worth funding for two or three years. This saves both parties time and expense.

2. No longer limit applications to postdoctoral fellows.
Impact—This broadens the scope of research to include practical studies by physical therapists, orthotists and/or nutritionists, etc. (non postdoctorals).

3. Create a strong collaboration between the CMTA, the National Institutes of Health, and the Medical Research Community to generate the resources to find a cure.
Impact—The massive amount of money necessary to fund important research can be realized through the National Institutes of Health and Federal Government. CMT research should get its fair share of NIH research dollars since CMT has an incidence of 1 in 2500 Americans. This will place the larger financial burden where it belongs, with the CMTA remaining as a catalyst for this initiative.

4. The CMTA must define the "case" or "product" of research that the development arm of the CMTA can utilize to solicit major financial gifts.
Impact—Experience has taught us that donors are more willing to give and give in a big way when we can present (define)

Medical Alert Listing to be Updated

The following medications are potentially toxic to CMT patients. Vincristine is proven to be hazardous to CMT patients, including those with no CMT symptoms, and should be avoided. However, all other listed medications are potential and not certain risks for worsening CMT neuropathy. All medication changes should be discussed with a treating physician. Drugs are grouped by relative risk.

Editor's note: This list has not yet been approved by the Board of the CMTA. It will replace the list found in all our publications once that approval takes place. It is proved here for you since it is the first major revision of the drug list in almost 15 years.

Definite high risk (including asymptomatic CMT):
Vinca alkaloids (Vincristine)

Moderate to significant risk:
Amiodarone (Cordarone)
Bortezomib (Velcade)
Cisplatin and Oxaliplatin
Colchicine (extended use)
Dapsone
Didanosine (ddI, Videx)
Dichloroacetate
Disulfiram (Antabuse)
Gold salts
Metronidazole/Misonidazole (extended use)

Moderate to significant risk (cont.):
Nitrofurantoin (Macrodantin, Furadantin, Macrobid)
Nitrous oxide (inhalation abuse or vitamin B12 deficiency)
Perhexiline (not used in US)
Pyridoxine (mega dose of Vitamin B6)
Stavudine (d4T, Zerit)
Suramin
Taxols (paclitaxel, docetaxel)
Thalidomide
Zalcitabine (ddC, Hivid)

Uncertain or minor risk:
5-Fluouracil
Adriamycin
Almitrine (not in US)
Chloroquine
Cytarabine (high dose)
Ethambutol
Etoposide (VP-16)
Fluoroquinolones
Gemcitabine
Griseofulvin
Hexamethylmelamine
Hydralazine
Ifosfamide
Isoniazid (INH)

Strategy

positively the research for which we request funding. Example: Dr. Zarife Sahenk, NT-3 Study at Ohio State University or Professor Michel Fontes, Ascorbic Acid Study, Marseille, France. These successes create enthusiasm, involvement, and commitment from donors.

The Board of Directors of the CMTA hopes to leverage our resources and utilize our influence to increase funding for research wherever possible.

When you receive your Appeal Letter, we ask that you consider our New Strategy and give as much as you can to this worthy effort. If you are interested in discussing a major gift to fund specific research initiatives, contact the CMTA at 1-800-606-2682. ✲

Uncertain or minor risk (cont.)
Mefloquine
Penicillamine
Phenytoin (Dilantin)
Podophyllin resin
Sertraline (Zoloft)
Statins
Tacrolimus (FK506, ProGraf)
Zimeldine (not in U.S.)
α-Interferon

Negligible or doubtful risk
Allopurinol
Amitriptyline
Chloramphenicol
Chlorprothixene
Cimetidine
Clioquinol
Clofibrate
Cyclosporin A
Enalapril
Glutethimide
Lithium
Phenelzine
Propafenone
Sulfonamides
Sulfasalazine

CMTA MEMBERSHIP/ORDER FORM

Name:_____

Address_____

_____

Phone Number: _____ Email: _____

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

	QTY	COST	TOTAL
Charcot-Marie-Tooth Disorders: A Handbook for Primary Care Physicians		*active members* $15 ***inactive members* $20**	
Membership Dues		$40	
CMT Facts I ☐ English ☐ Spanish		*active members* $3 ***inactive members* $5**	
CMT Facts II ☐ English ☐ 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**	
CMT Facts V		*active members* $12 ***inactive members* $15**	
A Guide About Genetics for the CMT Patient *No shipping and handling on this item only.*		*active members* $4 ***inactive members* $5**	
Golf Shirt *Size:* ☐ M ☐ L ☐ XL ☐ XXL		$15	
CMT Informational Brochure ☐ English ☐ Spanish		FREE	
Physician Referral List: States: _____ _____ _____		FREE	
Letter to Medical Professional with Drug List		FREE	
Contribution to CMT Research *10% will be applied to administrative expenses.*			
Shipping & Handling *Orders under $10 add $1.50, orders $10 and over add $4.50*			
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 or Fax to 610-499-9267.

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.

Ann Lee Beyer

1938-2004

On April 3, 2004, the Charcot-Marie-Tooth Association lost one of its earliest members and staunchest supporters when former Chairman and President, Ann Lee Beyer, lost her fight with cancer.

Ann was born Ann Marie Lee in New York City in 1938 to Irish immigrant parents. At the age of 1, Ann moved to Ireland with her parents and lived there for one year before catching one of the last boats out of Europe during the beginning of World War II. They returned to New York City and lived in the Washington Heights area. Ann attended St. Rose of Lima Grade School and graduated from the School of the Blessed Sacrament High School, both in NYC. After high school, she attended the City University of New York (CUNY) for one year before leaving her studies to help support her family.

Ann left Washington Heights when she married her husband, Ron Beyer in 1958. With Ron, she lived in Fairlawn, NJ; Savannah, GA; Monroe, LA; Ridgeway, PA; Park Ridge, NJ; and finally Upper Saddle River, NJ, where she remained for the rest of her life.

During the 1970's and 1980's Ann was affiliated with numerous church groups and ran several bible studies, youth groups, and adult spiritual groups. She was also actively involved in civic groups like the town-sponsored anti-drug campaigns of Park Ridge, NJ.

In 1983, Ann became affiliated with the fledgling organization, The National Foundation for Peroneal Muscular Atrophy, now called the Charcot-Marie-Tooth Association. She was an original support group leader for patients and families dealing with CMT and wrote the first edition of a pamphlet called "How to Start a Support Group."

In 1988, she wrote an article on "Families and CMT" which appeared in the The NFPMA Report (now The CMTA Report) and was a speaker at the Columbia University Symposium "Psychosocial Well-Being in Muscular Dystrophy and Allied Diseases III—Perspectives of the Future." She was then, and remained until her death, a close friend and associate of Dr. Robert Lovelace, the former Chairman of the CMTA's Medical Advisory Board.

Ann received her Bachelor of Arts degree in anthropology from Ramapo College and her Masters degree in anthropology

Over the years, Ann represented the CMTA at a booth at the American Neurological Assocation, the Human Genetics meetings, and the International CMT consortiums.

from Columbia University. She worked on a doctoral program in Anthropology at Columbia and traveled extensively in Ireland researching the traditions of the Irish wake. Her doctoral thesis focused on the works of James Joyce, especially *Finnegan's Wake* and the customs he of the Irish wake. She never completed her thesis because her focus changed to an emphasis on the involvement of her family and herself with the degenerative nature of CMT. Her interest in the CMTA became a priority in her life, and she devoted many hours to the work of the organization.

Ann was instrumental in the CMTA's involvement in the North American Database and the organizations' ties to the Wayne State CMT Clinic. Her passion was always research into the causes of CMT, the possible cures and the day to day function of CMT patients. She was a member of the CMTA's Board of Directors and finished her career with the CMT as both its President and Chairperson. Funding research grants was greatly increased during her tenure as Chairperson and the CMTA is now recognized as a leader in the field of CMT research due, in part, to her passion.

In recognition of Ann's contributions to CMT research, the plenary lecture at the International European CMT Consortium will be named the Ann Beyer Lecture. This will be the highlight of the July 8-10 meeting. Additionally, the CMTA's Board of Directors named a research grant the Ann Lee Beyer CMTA Research Fellowship. That grant was given this year to Dr. Yan Huang, who is working at the University of Pennsylvania under the mentorship of Dr. Steven Scherer.

Dick Sharpe, who served as interim Chairperson and President when Ann went on medical leave, said of her, "I remember meeting Ann at a patient/family conference in Florida about eight years ago. She was my first contact with the CMTA Board. I wanted to help and participate and within a year I had joined the Board and have remained active since then, largely because of my meeting with Ann. She will be missed by all of us who served with her on the Board over her fifteen-year period."

Executive Director Charles Hagins, who was hired by Ann, said that she always stressed three qualities she believed the CMTA should possess: professionalism, excellence, and integrity. Ann's favorite comment to me, he said, was "We must take this association to the next level of organization." Under her leadership, we have!

Finally, new Medical Advisory Board Chairman, Michael Shy, MD, wrote of Ann, "Ann's patience and calmness helped the CMTA Board survive in difficult times, and under Ann's leadership, the CMTA has evolved into a very important organization that now brings together leading scientists from all over the world and provides more information on CMT to patients than any other organization in the world."

Ann is survived by her husband, Ron, as well as five children and nine grandchildren. She will be missed by those who knew and loved her. ✲

CMT AND ASCORBIC ACID

(Continued from page 1)

action of ascorbic acid on PMP22 expression. However, several other targets are possible (we have another good candidate). Our goal is to determine as soon as possible if ascorbic acid works in CMT1A patients, the assumption being that if it does not work in CMT1A patients, it will not work in patients with other forms of CMT, meaning that ascorbic acid is only active in the animal model.

If it works in patients with CMT1A, then patients with other CMT forms should be tested. However, the dose might be different in different CMT subtypes.

Regarding doses and self-medication, it is perfectly understandable that CMT sufferers are eager to test the potential benefit of ascorbic acid. However, if patients self-medicate, we will never know if the molecule really works, because it must be tested in double-blinded fashion.

It would be most unfortunate if such a trial turned out to be impossible because of self-medication, because then ascorbic acid would never be recognized as a real medicine for CMT. One would never have data on possible toxicity. For instance, who can say that ascorbic acid at a high dose administered for 10 to 40 years is not toxic? Moreover, this drug would never obtain approval of national, European, or US agencies and patients may never get insurance coverage. The price of a daily high dose over decades is not insignificant. ✲

Ill Retiree to Test His Mettle by Turning Bicycle's Pedal

MIKE GOODWIN, PHOTOGRAPHS © STACEY LAUREN

SCHENECTADY - A neurological disorder has robbed Tony Palumbo of his ability to walk without leg braces. But it's not going to stop the 80-year-old Rotterdam retiree from pedaling 1,500 miles to raise money for an organization that helps troubled children.

Palumbo will fly to Jacksonville, Fla., today to begin a month-long solo journey to Schenectady on his 21-speed Trek bicycle to raise money for Northeast Parent & Child Society of Schenectady.

Fitted with special braces that give him the support he needs to pedal 60 to 100 miles a day, Palumbo is making his second multi-state bike ride to raise money for the agency.

"I've lost a lot of muscle mass because of this," Palumbo said of the Charcot-Marie-Tooth neuropathy that he has battled for several years.

Charcot-Marie-Tooth neuropathy is a hereditary ailment that leads to muscle weakness, atrophy and the loss of nerves and tendon use.

"The only good thing about it is it's slowly progressing," said Palumbo, who expects he will eventually need to use a wheelchair.

Nonetheless, he said he was anxious to get under way today on what he expects will be the last major bike ride of his life. He expects to end his trip in mid-June.

Tony Palumbo, 80, holds his Trek bicycle in Schenectady.

His love of the activity began in the 1940's when he and his 13-year-old son rode their bikes to Ausable Chasm in Essex County.

"He's incredible," said Laura B. Alpert, development coordinator for Northeast Parent & Child as she and Palumbo spoke recently.

"Let's wait until I finish the trip to say that," said Palumbo, who spent more than a decade as the organization's maintenance man after retiring from the Watervliet Arsenal.

In 1994, he biked from Vancouver, British Columbia, to Schenectady, a 3,200 mile trip that raised $21,000 for Northeast Parent & Child. This time, Palumbo's journey will help raise funds for the new Mohawk Dormitory Treatment Program, a residential treatment home for boys ages 12 to 16 who have been the victims of abuse and neglect.

It's the first time he has undertaken such a mission since he was diagnosed with Charcot-Marie-Tooth neuropathy. The ailment is slowly depriving him of the use of his muscles and has left him dependent on braces to walk.

Palumbo expects to start his ride Wednesday. He'll head north on U.S. 17 through the South before switching to U.S. 1 near the Mason-Dixon line. At nights, he plans to stay with friends or at roadside motels.

Palumbo is expected to telephone Don Weeks' morning radio show on WGY (810 AM) each morning to give progress reports. ✲

Exercise and CMT
Is It Worth the Effort ?

ROBERT A. BEDOTTO PT, CPO, www.ortho-therapy.com

Exercise implies effort and requires energy to perform. CMT requires you to budget your energy in order to maintain an active daily lifestyle. As activities become more difficult, more effort is required, thereby decreasing function. This is true for walking. Can exercise help if you have already exceeded your energy budget? Can exercise be detrimental? The answer to both questions is yes and no. The most important question is how to do it right. A more descriptive term for exercise might be movement therapy. The concept of movement therapy implies a goal-oriented approach to exercise. The ability to perform activities with the least amount of effort requires efficiency of movement patterns. The goal of movement therapy is to reduce the effort of the task being performed.

Exercise is often associated with muscle strengthening. Weights or other forms of resistance are commonly used. This does not necessarily lead to improved function or efficiency; in fact, it may be counterproductive. Muscles weakened by CMT should not be the focus of exercise. Overuse of these muscles in walking or other exercise can lead to fatigue and functional deterioration. The function of these weakened muscles in walking must be maintained through the use of an appropriate orthotic support system (brace). The normal muscles unaffected by CMT can become weak through disuse. Over the years, the weak muscles and resulting muscle imbalance cause compensations in the walking pattern; consequently, normal muscles weaken and function decreases as effort increases. These normal muscles can be strengthened. The focus should be on movement rather than resistive exercise.

Muscles weakened by CMT should not be the focus of exercise.

The primary concern in movement therapy is joint range of motion and muscle flexibility. Limitations in range of motion that are fixed are called contractures. Many so-called contractures are functional in nature. Tight muscles cause this type of limitation in range of motion. Any limitation in range of motion will cause functional deficiencies. In walking these deficiencies become exaggerated due to gravity and compensations for balance and security. This leads to increased effort and decreased efficiency. The emphasis should be placed upon improving the range of motion before strengthening is attempted.

Stretching also needs to be reviewed. As in the case of exercise, stretching is often done improperly. In order to increase muscle flexibility, the joint and muscle to be stretched must be positioned properly to avoid strains. Adjoining joints and related muscle groups must also be considered. The stretch should be minimal and sustained. It must be repeated routinely over a period of time in order to see results. Pain should not be experienced. Quick stretches and overstretching are counterproductive.

Stretching should be followed by movement of the body segment through the maximum range of motion available without resistance. Gravity must be considered. If the joint movement is affected by weak musculature, then gravity must be eliminated. The goal is to complete the movement. The movement can be assisted at first, followed by active movement in various positions. The body can create its own resistance by using the arms, legs, and trunk as levers. Gravity adds further resistance. Resistance should only be considered when the body segment can complete the full range of motion against gravity. The mere attempt to lift the body segment against gravity is excessive and fatiguing. This type of exercise will not make the muscle stronger and will only drain

(continued on page 10)

GIFTS WERE MADE

IN MEMORY OF

Irving Bank
Eileen Meltzer

Ann Lee Beyer
Mona & Laurence Beyer
Ed & Judie Butchko
Nancy & Martin Damiani
Fred & Carol Durr
Seth & Nancy Egelston
George Felter
Patrick Galizio
Victoria Haas
Michael & Jean Hennelly
Employees Kellogg Snacks
Robert & Pamela Kleinman
Bob & Carol Kobbe
Dan & Carol Leahy
John & Margaret Leahy
Sandra Leahy
Harry & Ilse Marasek
Frank & Adrienne Mellana
Sam & Leslie Mufson
Mim & Gary Reynolds
Cynthia Richter
Regie Roth
Phyllis Sanders
Alice & Bud Scott
Richard & Sheila Sharpe

Ann Lee Beyer (continued)
Judith Shipp
Michael & Rosemary Shy
Ronald & Nida Vallancourt
Ethel & Jack Walfish

Tiney Carroll
Joyce Wood

Max Ellis
Chris Dockstader
Wanda, Wayne, Ken, Brett Ellis

Mr. Gill
Brenda & Lee Hopkins

Guy Norton
Margaret Jean Smith

Stephen Paige
Timothy & Diana Dye
David & Arlene Fredricks
Barb & Mike Hall
Norman & Delight Inman
Mr. & Mrs. Alvin Kasson
Gary & Lorrie Katelman
Deana & Mike Liddy
Ron & Linda Lucke
Stanley & Irene Marcus
Jay & Bonnie Martindale
Phyllis Metzger
Delores & Douglas Miller

Stephen Paige (continued)
Karen & Russell Montgomery
Faye & Rocky Perrotta
Sheila & Fred Small

Rick Rogers
Edward & Louise Wilson

Pearl Ryan
Ralph & Deborah Militello

Roger VanderBoom
Kimberly Kiplinger & Peter Knowlton
Kathleen VanderBoom

William Wallace, Jr.
Gerald & JoAnne Adamson
Dean & Peggy Montgomery

Dean Weston
Darlene Weston
Nadine Zimmerlund

IN HONOR OF

Susan Chetlin & Michael Heydt
Charles & Marilyn Freed

Joe Gelman
Leon Gelman

Charles Ratcliffe, Jr.
Mr. & Mrs. Richard Davis

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: _____

_____

Other Ways to Support the CMTA

There are many ways that you can help support the important work of the CMTA. Here are just a few ways you can help:

- **Combined Federal Campaign (CFC) Giving:** This is the giving campaign of federal government employees. Our CFC number is 2513.
- **United Way:** Designate your United Way gifts to the CMTA when you are asked to give at your place of employment.
- **Employer Matching-Gift Programs:** Ask your employer to match your donation to CMTA.
- **Annual Research Drive:** Contribute annually to the CMT research drive in support of important research studies.
- **Shopping on the Web:** Shop at www.GreaterGood.com or www.iGive.com and for each purchase you make, a donation will be made to CMTA.
- **Annual Memberships:** There are individual and gift memberships available.
- **Stock Donations:** Donating appreciated securities, such as stock, to the CMTA.
- **Deferred Giving:** Remember the CMTA in your will.
- **CMTA Annual Fund:** Donations made to help support the operations of the CMTA.
- **Host Your Own Fund Raiser:** Contact the CMTA for guidelines and help.
- **Individual Donations:** Individuals can make donations in support of the CMTA's programs.
- **Support Groups:** Consider fund raising for the association.
- **Memorials/Honor:** Remembering and honoring family and friends. Instead of birthday presents for a major birthday, ask your family and friends to make donations to the CMTA in your honor.
- **Family Funds:** Families pooling their money together to support CMTA programs and research.

These are just a few ways you can help the CMTA in providing meaningful programs and promising research. The CMTA is a nonprofit, tax exempt 501(c)(3) organization. Donations made to the CMTA are tax deductible to the fullest extent of the law. ✲

Children Distribute Flowers in Memory of Marah Griffith

The 7th Annual "Daffodils for Seniors" took place Saturday April 3, at Laurel View Village, Johnstown, PA. Children pick, arrange, and distribute thousands of daffodils at no cost to residents of nursing homes every spring.

The daffodil giveaway is done in the name of Charcot-Marie-Tooth Disease (CMT) and in memory of Marah Griffith, who had CMT and died at the age of sixteen on December 25, 2001. Marah was instrumental in the concept of children giving daffodils to seniors. Marah loved organizing the event and working with the children. ✲

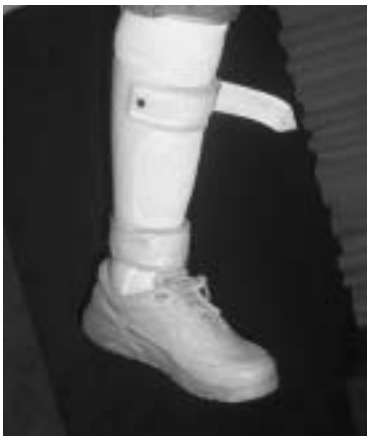
SUPPORT GROUP NEWS

Pennsylvania – Johnstown Area

The Johnstown Support group met April 24th at the Myron Williams Conference Room at Conemaugh Health System. The featured speaker, Jason Cook, PsyD, gave an excellent talk on the role of worry and stress on health and, in particular, CMT. The techniques he demonstrated were very useful and he had us meditating by the end of the presentation. JD gave a talk on current research, and a recent study from France on ascorbic acid helping CMT-1A transgenic mice got the group excited (one of the researchers received support from the CMTA). The group also talked about the upcoming conference where Dr. Shy will be presenting in Johnstown.

Pennsylvania – Pittsburgh

The Pittsburgh CMTA Support group held its initial meeting Saturday May 8th at the West Wing Auditorium at the University of Pittsburgh, Shadyside. The meeting was organized by JD Griffith of the Johnstown area support group. He gave a presentation on CMT and recent research into the disease. When JD couldn't get the projector to work, he had everyone gather around the laptop, which made for a more intimate meeting. Bracing was the most discussed topic with lots of show and tell. The group decided to include a bracing expert in the next meeting program. If you want to join or help, please contact: Janet Fierst, 412-761-0910, E-mail: jfierst@att.net or JD Griffith, 814-539-2341, E-mail: jdgriffith@charter.net ✲



"Show and tell" in Pittsburgh involved patients showing the braces they use to improve their gait.

EXERCISE AND CMT

(Continued from page 7)

valuable energy that could be put to better use.

The goal of movement therapy in rehabilitation is to improve function. Walking efficiently is the best exercise possible. In order to accomplish this goal, weak muscles must be supported, balance must be restored, and normal muscles must be allowed to function appropriately. The use of an appropriate brace is often indicated. The long-term result should be to reduce effort and save energy rather than deplete it.

To the average individual, exercise and stretching seem simple. In actuality, it is a very complex science. Highly trained athletes rely on the expertise of physical therapists and trainers who incorporate biomechanical principles into exercise programs. They function at the highest level of efficiency. Ask any athlete about stretching and he or she will tell you how important it truly is. While you do not want to engage in high-level athletic events, your activities of daily living can be even more demanding!

Movement therapy is about movement. It does not focus on strengthening or exercise. Even limited ambulators or non-ambulators need movement therapy. Cardiovascular fitness is of concern for all of us! Swimming and mat activities are good examples of movement therapy that increase flexibility and endurance without excessive resistance or fatigue. A vicious cycle of increased effort and reduced function begins as lifestyles become more sedentary.

Movement is a necessary part of life. Our physical and psychological well-being depends on it. The question should not be whether or not to exercise but how to do it right! Stop exercising and start moving! Activities that require less effort will be more enjoyable and promote activity rather than discourage it. Improper exercise and stretching can be detrimental, but not being active is worse! Activity must be maintained. Movement therapy must become a lifestyle choice. Learn how to do it right!…… MOVE…. for LIFE! ✲

CMT Support Groups

Bob Budde, Support Group Liaison, 859-255-7471

Arkansas—Northwest Area
Place: Varies, Call for locations
Meeting: Quarterly. Meetings are not regularly scheduled so call ahead.
Contact: Libby Bond, 479-787-6115
Email: charnicoma57@yahoo.com

California—Berkeley Area
Place: Albany Library, Albany, CA
Meeting: Quarterly
Contact: Gail Whitehouse
Email: gwhite@earthlink.net

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
Email: pcmobley@mac.com

Colorado—Denver Area
Place: Glory of God Lutheran Church Wheat Ridge
Meeting: Quarterly
Contact: Marilyn Munn Strand, 303-403-8318
Email: mmstrand@aol.com

Kentucky/Southern Indiana/ Southern Ohio
Place: Lexington Public Library, Northside Branch
Meeting: Quarterly
Contact: Martha Hall, 502-695-3338
Email: marteye@mis.net

Massachusetts—Boston Area
Place: Lahey-Hitchcock Clinic, Burlington, MA
Meeting: Call for schedule
Contact: David Prince, 978-667-9008
Email: baseball@ma.ultranet.com

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

Minnesota—Twin Cities
Place: Call for location
Meeting: Quarterly
Contact: Maureen Horton, 651-690-2709
Bill Miller, 763-560-6654
Email: mphorton@qwest.net, wmiller7@msn.com

Mississippi/Louisiana
Place: Baptist Healthplex, 102 Clinton Parkway, Clinton, MS
Meeting: Quarterly
Contact: Flora Jones, 601-825-2258
Email: flojo4@aol.com

Missouri—St. Louis Area
Place: Saint Louis University Hospital
Meeting: Quarterly
Contact: Carole Haislip, 314-644-1664
Email: c.haislip@att.net

New York—Greater New York
Place: NYU Medical Center/ Rusk Institute, 400 E. 34th St.
Meeting: Third Saturday of each month from 1-3 PM of each month
Contact: Dr. David Younger, 212-535-4314, Fax 212-535-6392
Website: www.cmtnyc.org

New York—Horseheads
Place: Horseheads Free Library on Main Street, Horseheads, NY
Meeting: Quarterly
Contact: Angela Piersimoni, 607-562-8823

New York (Westchester County)/ Connecticut (Fairfield)
Place: Blythedale Hospital
Meeting: 3rd Saturday of each month, excluding July & August
Contacts: Beverly Wurzel, 845-783-2815
Eileen Spell, 201-447-2183
Email: cranomat@frontiernet.net espell@optonline.net

North Carolina—Archdale/Triad
Place: Archdale Public Library
Meeting: Quarterly
Contact: Ellen (Nora) Burrow, 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
Email: Greenville-Ohio-CMT@woh.rr.com

Ohio—NW Ohio
Place: Medical College of Ohio
Meeting: Quarterly
Contact: Jay Budde, 419-445-2123 (evenings)
Email: jbudde@fm-bank.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
Email: jeanie4211@hotmail.com or blzerbabe@aol.com

Pennsylvania—Johnstown Area
Place: Crichton Center for Advanced Rehabilitation
Meeting: Bimonthly
Contact: J. D. Griffith, 814-539-2341
Email: jdgriffith@mail.charter.net

Pennsylvania—Northwestern Area
Place: Blasco Memorial Library
Meeting: Call for information
Contact: Joyce Steinkamp, 814-833-8495
Email: joyceanns@adelphia.net

Pennsylvania—Philadelphia Area
Place: Penn Towers Hotel Conference Room
Meeting: Bimonthly
Contact: Amanda Young, 215-222-6513
Email: stary1@bellatlantic.net

Pennsylvania—State College
Place: Centre County Senior Center
Meeting: Monthly
Contact: Rosalie Bryant
Email: rab296@psu.edu

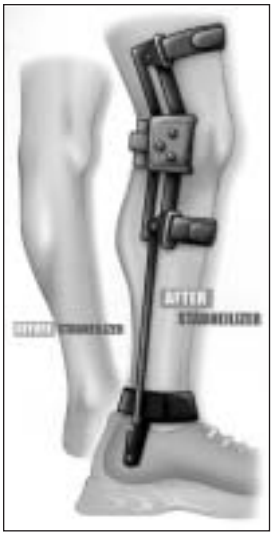
Another Alternative in the Bracing of CMT

A member of the CMTA sent the following letter to the newsletter:

I am a longtime subscriber to the report, but, this is my first time writing to the association. I am 52 years old and have had CMT all my life. At age seven, my parents were told by a doctor that both my older brother and I had muscular dystrophy. It wasn't until I was 27 that I was finally told it was CMT. I was able to have a successful career in retail management that lasted until age fifty. At that time, due to the progression of my CMT and another illness, I had to retire. But the reason I'm writing is not to tell you about me, but to introduce you and your readers to a wonderful man I met almost six years ago. It was at that time I attempted to get fitted for AFOs. I met with an orthotist who put the rigid braces on my legs. I knew immediately these would never work for me. I had been falling regularly due to dropfoot. After exhausting all of the alternatives, I was extremely depressed and ready to leave. It was then that the orthotist pulled out a device called The STABHEILIZER. He told me a story about a man named Dean Heil, a Houstonian and a fellow CMT patient, who had invented the brace he was about to show me.

I put on the STABHEILIZER and it was as if a miracle had occurred. Not only did it eliminate my foot drop, but it stabilized the ankles and allowed me to walk with confidence. I can't begin to tell you how thrilled I was. The STABHEILIZER allowed me to continue in my job for the next five years. Without it, I surely would have had to retire much earlier.

It was at this time, not ever having met someone with CMT, that I was determined to meet Dean. Arriving at his home one afternoon, I met a quiet, unassuming man who possessed a single-minded desire to help those affected by CMT. He had owned his own construction company in Houston for over 30 years. After a terrible saw accident on the job, he was laid up with nothing to do. It was then that he became determined to construct a bracing device that would accommodate those people still active enough that traditional AFOs were not an option. After years of working out of his garage, he developed a light-weight and strong, easy-to-use product which produces the most natural gait I've ever seen.



The STABHEILIZER

Your report has been my number one source for information concerning CMT. Dean is now in the process of marketing the STABHEILIZER to the orthotic community. I would love for you and the CMT community to know about Dean and his invention.

The patented STABHEILIZER is light-weight, consisting of Velcro straps anchored just above the ankle joint. Two lateral metallic strips extend from the anchored strap to the outside of the shoe. Anteriorly, one strap extends from the anchored strap to the inside of the shoe attached to the tongue of the shoe. The amount of dorisflexion can be controlled with this anterior strap. Donning of the brace is easy and accommodates changes in girth/growth of extremity without alteration or discomfort. The brace is useful for hypotonic or atonic musculature of the lower leg. However, severe spastic footdrop and contractures cannot be managed with the STABHEILIZER.

The STABHEILIZER offers both hyperextension support and foot lift. With the extension bar attached to the outside of the shoe and the metallic strip of the STABHEILIZER, dorsiflexion of the ankle is achieved and at the same time, the knee is prevented from hyperextending. These devices are approved by Medicare and may be covered by your insurance.

You can view Dean's products on the website at www.stabheilizer.com or you may speak to Dean at 281-580-0555.

—John Dineen, Texas

Editor's note: As with all devices, this will not provide adequate support for many CMT patients and should be evaluated by your podiatrist, orthopaedist, or orthotist before purchase.

ASK THE DOCTOR

Dear Doctor,
I am taking Bactrim for a UTI. Is it a sulfa drug? I have had CMT since early childhood. Now, I am having much more weakness when transferring from my wheelchair. My hands are very affected and my breathing, with phrenic and larynx involvement, has gotten worse. I also take Celexa and I read about SSRIs and CMT. Do you think this is a problem?

The Doctor replies:
Bactrim is a sulfa-containing drug. There is a very rare and not generally accepted link with neuropathy and Bactrim. Considering the millions of doses given, the link is probably not valid. There is no specific knowledge of CMT patients getting worse on Bactrim and the drug was removed from the soon-to-be-released updated drug list for the CMTA. (*See page 2 of this issue*). SSRI drugs have been under suspicion, but not Celexa in particular.

Dear Doctor:
I am a 47-year-old female with CMT1A. When I was 15 years old I had a triple arthrodesis on both ankles. I recently had another ankle fusion on my left ankle because the joint had deteriorated and was arthritic. The problem is the bone is not regenerating quickly enough and at present one screw has broken and another is bending. There are five screws altogether. I might also add the surgery was performed Nov. 17, 2003, so it is 5 1/2 months already.

My question—is there anything I can take or do, to help in the regeneration of the bone? I am obviously desperate to try and find some solution or it's back to surgery.

I would appreciate any information you can give me on this problem.

The Doctor replies:
Let me try to sort things out a bit. Triple arthrodesis, as we know it today, is not an operation on the ankle. It is actually an operation on the hind-foot, fusing together the talo-calcaneal joint; the talo-navicular joint; and the calcaneal-cuboid joint (thus triple = 3 arthrodesis = joint fusion). It is an operation that would help correct a foot that is deformed after growth has ceased. In this case, it was done 32 years ago.

During these 32 years, because these joints no longer move, there is added stress on the ankle joints and the ankle joints then deteriorate and become arthritic. That would cause swelling and pain and thus, it would be reasonable to propose the fusion of that joint.

Ankle fusions are sometimes technically very difficult to do and to maintain, not only in persons with CMT disease, but also in people who have had accidents or damage to the ankle joint and would need to have their ankles fused. If the screws used to hold the fusion are bending and breaking, then there is motion in the area that is to be fused and that is not good. There is nothing you should do, take, etc., as you need to continue medical treatment, but there is something that your surgeon should do as part of his/her responsibility to your care, and that is to continue caring for the problem.

Although bone healing does depend a lot on the nature of the quality of bone, immobilization may be necessary. Immobilization by the use of the five screws was not sufficient and thus, there was motion, causing the breakage of the screws. The orthopaedist should consider whether there is need for revision and correction to attain the the intended fused position, or whether cast immobilization and orthosis would help temporarily to hold the position and allow the fusion to become more mature and "solid."

Disuse leads to osteoporosis—that is a totally different matter, but it needs to be considered by the surgeon who can incorporate that aspect into the total care of the ankle fusion. To help bone heal, sometimes electromagnetic bone stimulators are helpful.

Dear Doctor,
Is it possible that SIDS (sudden infant death syndrome) is an early form of CMT?

The Doctor replies;
SIDS is not a disorder of the peripheral nervous system. The peripheral nervous system, which is *always* involved in CMT is not affected in SIDS. ✳

LETTERS

WRITE TO US!

Pat Dreibelbis, Editor
The CMTA Report
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Chester, PA 19013
or
CMTAssoc@aol.com

The CMTA reserves the right to edit letters for space.

Dear Pat/CMTA:

I just read the last newsletter which was sent to Rancho and since I am "semi-retired," I do not go to the Chief's office (my old office) very often. I am sorry to hear about your accident, but it looks like you are doing OK and you haven't missed too much work (which you are supposed to do).

I also broke my right shoulder in March, 2003 in Australia when I did not notice and fell from a step and had to get treatment on a Sunday morning at the public hospital in Sydney. I had to have it immobilized for 3 weeks until it healed sufficiently. It was on the socket side and not displaced enough to need surgery. I had to learn while traveling with my wife for the next 2 weeks in Australia to be one-handed, not to use it, and be patient so that it would heal. Then I started to "rehabilitate" myself by exercises, use, functional training, etc. (following all the steps and making sure all the details were followed, which I make my patients do); I went through the same experience you went through and the fracture healed well. I have no motion loss, limitation, or disability. I returned to swimming and lifting weights.

I trust that everything is getting back to normal. Keep up the good work. Both of us have learned a lesson, not only of humility, but of what our patients go through all the time. That is why we listen to them and why what they tell us about their problems and needs is very important.

—Best wishes,
John Hsu, M.D.

Dear CMTA:

Fifty years ago I just needed a traditional toe-lift AFO so I could walk with shoes on. I've visited various orthotists over the years and although I'm not medically trained, I've come to believe that too many orthotists don't recognize the need for more support, as the leg muscles deteriorate with age in CMT patients. The typical toe-lift AFO is lightweight and is designed to simply lift the toes when walking. But, if a patient has developed dropfoot, due to say a stroke, the remaining leg muscles are still normal strength. Which is apparently why amputees can ski and run with artificial legs while most CMT patients can't, since they don't have the other supporting leg muscles.

In my 70's I could no longer stand without holding on to something. Fortunately, my San Diego orthotist works with a great many CMT patients and recognizes the need for greater support to make up for the progressive wasting of the leg muscles. My AFOs are now sturdier (thicker material) and encompass more of my calf and have a sturdy leg strap. I can now stand without holding on to something. My layman's analysis is that the added AFO calf support makes that possible.

My point in writing is that, too often, until something hits us between the eyes (so to speak) we don't realize that there's a possible solution to our prob-

New York Support Group Hosts Regional Meeting

Dr. Michael Shy

Saturday, June 19, 2004, Dr. David Younger and the Greater New York Support Group will host Dr. Michael Shy. His keynote address will be entitled, "Therapeutic Advances in Charcot-Marie-Tooth Disease."

The day begins with a buffet luncheon at noon, followed by Dr. Shy's lecture.

The meeting will be held in the Faculty Dining Room of the New York Medical Center, 550 First Avenue at 32nd Street, New York, NY. Questions? Call Dr. David Younger at 212-535-4314.

lems. Maybe this will help someone with problems similar to mine.

By the way, I also find that with my currently added AFO support, it is increasingly difficult to walk around the house without my AFOs on and I'm told this is a frequent result.

I don't know about you, but the first thing I do when I come home is to take the AFOs off and remove my hearing aid and glasses. Which reminds me of a very old joke: the man, talking to his advice analyst, says his wife at night takes off her wooden leg and puts it in a drawer followed by her glass eye, hearing aid, and her wig. His question is: should he sleep in bed or in the drawer?

I'm so grateful my wife doesn't opt for the drawer.

—Bob via e-mail

Dear CMTA:

In my article in the Fall 2003 newsletter called "Things that work for me" I recommended a water/beach shoe, but lamented the loss of a retail source. I had worn a hole in the sole of my last pair when I found them in the L.L. Bean Catalogue. Bean calls them "Bean's shoreline water shoes" and the stock numbers are RT47910(Men's) and RT49049(Women's). I ordered a pair and a spare!

—Jack Veatch

Dear CMTA:

I am a 74-year-old male with CMT1A. Until now, I have been pretty successful at dealing with the obstacles CMT has presented. However, I now have a problem with which I could use some help.

As a result of my CMT, I do not have the hand strength to spread and pull my stockings onto a sock puller. I wear long stockings between my legs and my AFOs, and mid-calf stockings over the AFOs for stability as well as cosmetics. I have found stockings without elastic tops, but I still cannot manage to do the task in less than 40 to 60 minutes using a pliers (which is pretty tough on the stockings). With my wife's help, the task is done in minutes; but she has her own health problems, so I need to strive for more independence.

Surely someone has come up with a device or procedure for getting stockings onto a sock puller and then up the leg for those of us with minimal hand strength.

—Hopeful

Saving Money on Your Healthcare Expenses

People with disabilities are often overburdened with medical expenses. Here are a few ways they may be able to save money:

- If your annual medical expenses amount to approximately 7.5 percent or more of your annual income, your healthcare bills may be tax deductible if you file the IRS long form with itemized deductions. Check with your tax preparer or phone the IRS at 1-800-829-1040.
- If you have retirement savings in an IRA or 401(K) and you become permanently disabled, you may be able to withdraw money from these accounts without a tax penalty even if you are not old enough to retire. Check with the managers of your retirement fund or your bank to obtain the rules that allow disabled people to make early withdrawals without penalty.

- Are you a veteran? If you served in the military, you may be eligible for healthcare services and pharmaceuticals from the Veterans Administration (VA). Few people realize that the VA runs the largest mail-order prescription drug service in the United States. If you are accepted into a VA program, you may be able to get prescription medications for a nominal fee.

However, each VA region has a drug "formulary," which means they provide certain drugs but not all brands of pharmaceuticals. Check with your local VA hospital or Veterans Administration office to find out how to quality. ✧

The CMTA Report is published by the Charcot-Marie-Tooth Association, a registered non-profit 501(C)(3) health organization. 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.

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).
- does not affect life expectancy.
- 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).
- 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, 1D (EGR2), 1X, HNPP, 2E, 4E, and 4F can now be diagnosed by a blood test.
- is the focus of significant genetic research, bringing us closer to solving the CMT enigma.

THE CMTA *Report*

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