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THE CMTA Report

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

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

CMTA Lobbies Congress on Capitol Hill

CHARLES F. HAGINS, Executive Director

> "Our efforts in Washington, DC, seem promising. We made a number of good contacts with various legislators who appeared sympathetic and promised to help the CMTA in its efforts to get more serious funding from the NIH. It is now important for the association and its members to follow up and contact their Congressmen to get on board with this effort."
>
> **—Robert Kleinman, CMTA Board Member**

On February 25, 2004, the CMTA Board of Directors held their quarterly meeting at the Longworth Building directly connected to the Nation's Capitol in Washington, DC.

The Board began their day by conducting the CMTA's necessary business agenda, followed by a full day's itinerary on Capitol Hill.

Jason Steinbaum, Chief of Staff for Congressman Eliot Engel (D-NY) organized the day on behalf of the Association. He began with a presentation on "How to Lobby Members of Congress." Next, Diane Blagman of Greenberg Taurig, former Associate Staff House Appropriations Committee and Chief of Staff for an Appropriations Subcommittee Chair, lectured the Board on "How to Pursue Appropriations."

The final session before lunch was conducted by Chuck Konigsberg, former Executive Director of the Parkinson's Action Network, who is currently Vice President for Public Policy of the United Jewish Communities, and who instructed the group on "How to Pursue NIH Grants."

The Board was challenged to create a single message or request to convey to Members of Congress on behalf of the Association with regard to advancing our cause. The message we agreed upon was to have Congress support joint resolution language in the fiscal year 2005 appropriations budget requesting the National Institutes of Health (NIH) to create a needs assessment survey with regard to current funding of CMT research.

With this message in hand, Board Members began visiting Members of Congress to convey the request.

Board Members Robert Kleinman and Phyllis Sanders called upon Representative Steve Israel (D-NY) to begin the process.

Lunch was held in H-139 Capitol Building with Representative Eliot Engel. Congressman Engel agreed to support our efforts and he informed the Board on how the President's tax cut and the war in Iraq will affect appropriations. The good news is the CMTA is not asking

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INSIDE:

Appeals and Membership—Some Clarifications

FROM THE CMTA EXECUTIVE DIRECTOR

As the CMTA moves forward in its lobbying of Congressmen and legislators for funding, our membership numbers are more important than ever. We need more members because our strength is in our numbers.

We begin this article with our thanks to those of you who supported our Annual Appeal in December, 2003 and subsequently. Your generosity has resulted in a total, so far, of $78,957. If you haven't given to the Annual Appeal, it's not too late, and your gift will be gratefully acknowledged.

Now for the clarification. We have just completed mailing approximately 900 postcards to people who paid their membership for the year 2002–2003 but failed to renew for this year. It has been our policy, in the past, to "carry" people for a few months and to continue sending them our newsletter even if they failed to renew their membership in a timely fashion. This is no longer possible because of the costs involved. So, in addition to the regular reminder of membership renewal, a blue postcard will now be sent to members who fail to renew within their monthly period.

The confusion seems to arise from a misunderstanding of how gifts to the CMTA are allocated. We have two fundraisers per year: one for the operating fund, called the Annual Appeal, and one for the research fund, called the Research Appeal. Gifts to either of these appeals are not considered membership dues because they are "gifts" and can be used as charitable contributions on income taxes. When a person returns an appeal gift in a labeled appeal envelope, the accounting staff assumes the contribution is intended as a gift to that appeal. Unless someone writes "for membership," the money will go into the other funds, and membership will still be due on the one-year anniversary of the member's last contribution to membership.

CMT Conference to be Held in Johnstown, PA

The Cambria-Somerset Council for Education for Health Professionals will present a one-day program called "CMT: New Awareness and Current Concepts for Health Care Providers, Patients, and Families" on Wednesday, May 26, 2004, at the Holiday Inn in Johnstown, PA.

The conference will begin at 7:30 a.m. with registration, refreshments, and visits to the exhibits and booths of display items. The welcome and introduction of the speakers will occur at 7:55, with the program officially beginning with Dr. Michael Shy of Wayne State University providing an overview of CMT. After a brief break, Dr. Neil Busis will discuss the diagnosis of CMT and Dr. Stephen Conti will talk about the CMT foot and surgical options.

Lunch will be provided from 11:25–12:25. Following the lunch break, Dr. David Riley will discuss CAM and CMT and Dr. Robert Chetlin will present his findings on CMT exercise research. The final presentation will be by Susan

Hopefully, members will understand this explanation. A gift to research is a restricted gift and cannot be used for anything other than research grants and symposiums. A payment of membership gives the person a subscription to the six issues of the newsletter, as well as discounts on all the publications we offer and any patient/family conferences we sponsor. A gift to the Annual Appeal helps fund our 800 number and our website. It also funds the mailing of informational packets to new inquirers, our attendance at medical conventions to inform medical professionals about CMT, and the printing of our *Facts* series and our *Physicians' Handbook*.

As always, we appreciate deeply the contributions and membership dues that our constituents are so faithful in sending us. We are only as strong as you enable us to be, and our goal is to be strong enough to ultimately find the cure for CMT. ✳

Wheeler of the Canadian publication, *CMT...Today*, who will talk about living with CMT.

Health care professionals can receive continuing education credits for attendance at the conference. Their costs range from $48-$60. The registration deadline is May 15, 2004, and the cost is $20 for patients, $30 for family members, and $40 for a one-year CMTA membership plus admission. Questions about this conference should be addressed to the Cambria-Somerset Council at 1-814-269-2978. ✳

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.

LOBBYING WASHINGTON

(Continued from page 1)

At lunch, Congressman Eliot Engel (D-NY) discussed his support of the CMTA's efforts in lobbying members of congress.

for new money, but a greater piece of the pie for CMT research.

Mary Jo Hocksema, formerly with NIH's Congressional Affairs and currently Public Affairs Specialist for the Population Association of America, discussed NIH's process for disease research requests.

> "Thanks to Jason Steinbaum's efforts, myself and other board members had the opportunity to meet with the United States Legislature on February 25, 2004. We informed our representatives about CMT and the importance of research dollars in finding a cure. We hope that this endeavor was a successful start in educating others about this crippling nerve disease and the immediate need to further fund research."
>
> **—Steve O'Donnell**

> "I never thought of myself as a lobbyist, but lo and behold, we, the CMTA Board and Executive Director became lobbyists on behalf of the CMTA when we met in Washington, DC, on February 25, 2004. I must say that the day was long and exhausting, but very beneficial for the organization. We had the opportunity to meet with various congressional people and to have lunch with our host, Jason Steinbaum and his boss, Congressman Engle. The question is: were the meetings a success? My opinion is that they were a great success. The seed has been planted about CMT and what our needs and goals are. I'm convinced our efforts will bring forth a greater awareness of our disease throughout the country and also provide us with a greater opportunity to obtain additional funding through NIH for research. I truly believe each and every member of this association can help our cause by writing or calling their local Congressman or Senator, telling them about CMT. Give it a try...it can only help."
>
> **—Patrick Torchia, Co-Chairman and Co-President**

The afternoon schedule consisted of calling on Congressional offices. Board Co-chairmen/Co-Presidents Patrick Torchia and Steve O'Donnell, Medical Advisory Board Member Dr. Michael Shy, and Executive Director Charles F. Hagins called upon Representative John P. Murtha (D-PA) and Representative Joseph Knollenberg (R-MI).

Phyllis Sanders and Robert Kleinman finished out a busy day by calling on Representative Carolyn McCarthy (D-NY).

If you have never been to the Capitol Building and surrounding campus, our members of the Board will tell you that you will walk miles of halls, corridors and connecting passages, some underground.

Everyone was exhausted by day's end; however, everyone shared a feeling of accomplishment.

Mission accomplished? No. *Just the first step!* But it was a huge step toward promoting awareness among our nations' leaders in the hope of generating research dollars for CMT at the NIH. ✳

Diane Blagman told the CMTA's Board of Directors how to pursue budget appropriations successfully.

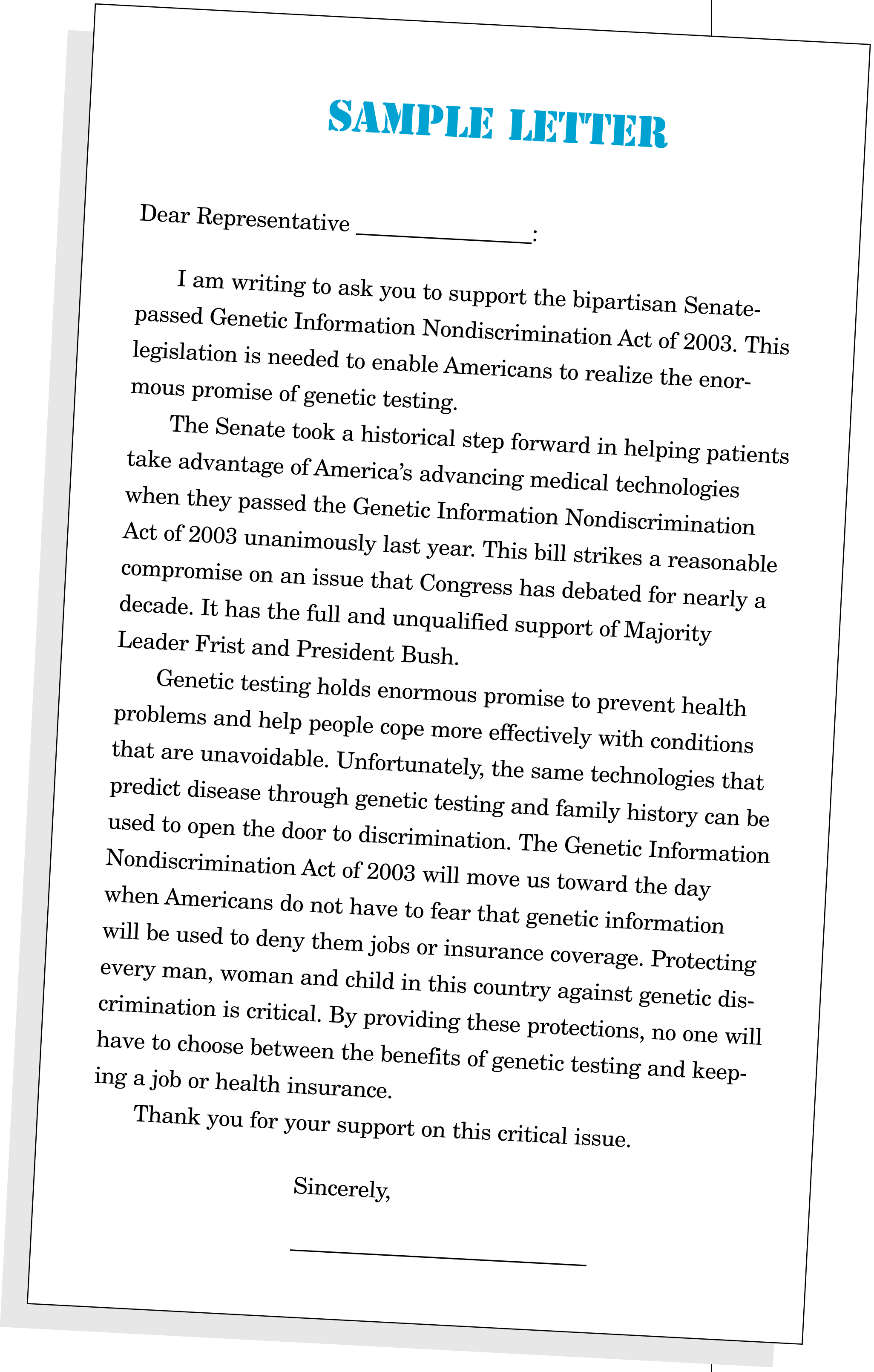
Genetic Nondiscrimination Action Alert

FROM NORD

The Senate took a historic step forward in helping patients take advantage of America's advancing medical technologies when they passed the Genetic Information Nondiscrimination Act of 2003. This measure is a significant advance that will move us toward the day when Americans do not have to fear that genetic information will be used to deny them job or insurance coverage.

Protecting every man, woman and child in this country against genetic discrimination is critical. Genetic testing holds enormous promise to prevent health problems and help people cope more effectively with conditions that are unavoidable. Unfortunately, the same technologies that predict disease through genetic testing and family history can be used to open the door to discrimination. This bill gives protection to Americans who fear that their genetic information could be misused to prevent them from getting the jobs or coverage they need. By providing these protections, no one will have to choose between the benefits of genetic testing and keeping a job or health insurance.

Please write to your representatives to convince them to pass this measure very soon, and encourage President Bush to sign it into law. We hope this victory will be the first in a line of measures that allow Americans to realize the enormous promise of genetic testing.



SAMPLE LETTER

Dear Representative ______________:

I am writing to ask you to support the bipartisan Senate-passed Genetic Information Nondiscrimination Act of 2003. This legislation is needed to enable Americans to realize the enormous promise of genetic testing.

The Senate took a historical step forward in helping patients take advantage of America's advancing medical technologies when they passed the Genetic Information Nondiscrimination Act of 2003 unanimously last year. This bill strikes a reasonable compromise on an issue that Congress has debated for nearly a decade. It has the full and unqualified support of Majority Leader Frist and President Bush.

Genetic testing holds enormous promise to prevent health problems and help people cope more effectively with conditions that are unavoidable. Unfortunately, the same technologies that predict disease through genetic testing and family history can be used to open the door to discrimination. The Genetic Information Nondiscrimination Act of 2003 will move us toward the day when Americans do not have to fear that genetic information will be used to deny them jobs or insurance coverage. Protecting every man, woman and child in this country against genetic discrimination is critical. By providing these protections, no one will have to choose between the benefits of genetic testing and keeping a job or health insurance.

Thank you for your support on this critical issue.

Sincerely,

____

CLINICAL TRIALS

Linking Patients to Medical Research

CLINICAL AND MOLECULAR MANIFESTATIONS OF INHERITED NEUROLOGICAL DISORDERS

Sponsored by National Institute of Neurological Disorders and Stroke (NINDS)

■ PURPOSE:

This study is designed to learn more about the natural history of inherited neurological disorders and the role of heredity in their development. It will examine the genetics, symptoms, disease progression, treatment, and psychological and behavioral impact of diseases in the following categories: hereditary peripheral neuropathies; hereditary myopathies; muscular dystrophies; hereditary motor neuron disorders; mitochondrial myopathies; ataxias; hereditary neurocognitive disorders; inherited neurological disorders without known diagnosis; and others. Many of these diseases, which affect the brain, spinal cord, muscles, and nerves, are rare and poorly understood.

Children and adults of all ages with various inherited neurological disorders may be eligible for this study. Participants will undergo a detailed medical and family history, and a family tree will be drawn. They will also have a physical and neurological examination that may include blood test and urine tests, an EEG (brain wave recordings), psychological tests, and speech and language and rehabilitation evaluations. A blood sample or skin biopsy may be taken for genetic testing. Depending on the individual patient's symptoms, imaging tests such as X-rays, CT or MRI scans, and muscle and nerve testing may also be done.

Information from this study may provide a better understanding of the genetic underpinnings of these disorders, contributing to improved diagnosis, treatment, and genetic counseling, and perhaps leading to additional studies in these areas.

■ FURTHER STUDY DETAILS:

Individuals and their families affected by neurological disorders with a potential genetic basis will be the focus of this screening and repository protocol. The goals of the protocol will be to contribute to both the clinical and molecular understanding of inherited neurological disorders. Clinical issues to be addressed will include disease manifestations, natural history, management and psychological/behavioral impact of disease. Patients with diseases of known molecular basis will be genotyped in order to investigate phenotype-genotype correlation. Patients with disease of unknown or incomplete genetic characterization will be studied with hopes of contributing to the identification of specific genes responsible for disease. It is anticipated that additional protocols will be generated from preliminary data gathered in this study. The study will also facilitate the training of fellows and students in neurology, neuroscience, and genetics.

■ INCLUSION CRITERIA:

Patients of all ages and their families with known or suspected inherited neurological disorders.

Pregnant women will be included except for tests involving radiation or anesthesia.

General health and well being of each potential participant must be sufficient to allow for travel to the NIH, blood drawing, and, as indicated, skin or muscle biopsy, pulmonary and cardiac evaluations, physical therapy assessments, and MRI.

Must be able to return to the NIH at least twice a year on a regular basis.

■ ENROLLMENT: 750

DIAGNOSTIC EVALUATION OF PATIENTS WITH NEUROMUSCULAR DISEASES

Sponsored by National Institute of Neurological Disorders and Stroke (NINDS)

■ PURPOSE:

This protocol has three purposes: 1) to screen patients with neuromuscular diseases for participation in NIND's Neuromuscular Diseases Section (NDS) research studies, 2) to train NDS fellows in evaluation of difficult neuromuscular cases, and 3) to provide follow-up for patients who have finished participating in a previous study and not enrolled in another. This study will not use investigational treatments.

Patients who 1) have a neuromuscular disorder suitable for a current or future protocol, 2) present a diagnostic puzzle, or 3) have an unusual manifestation of a more usual neuromuscular condition may be eligible for this study. Neuromuscular disorders may include peripheral neuropathies, myopathies, dystrophies, mayasthenia, stiff person syndrome, muscle cramps and pains, myotonias, motor neuron diseases, and post-polio syndrome.

Participants may undergo the following diagnostic studies:

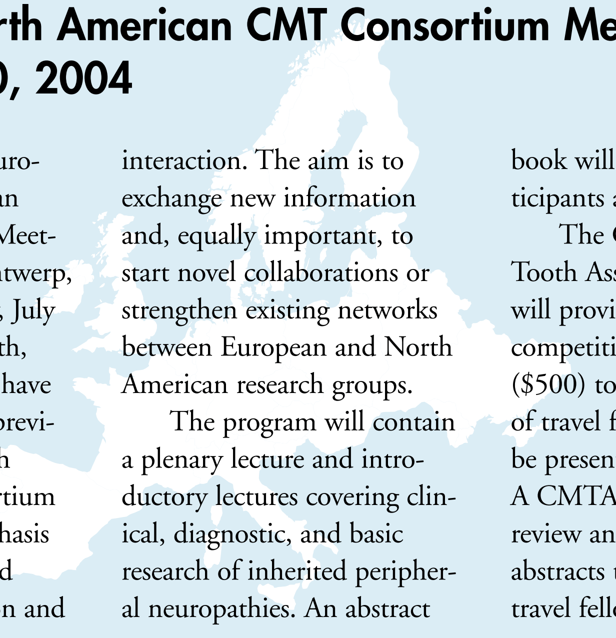
- *Blood drawing*—No more than 100 ml (less than 4 ounces) of blood will be drawn.
- *Electromyography (EMG) and nerve conduction studies*—EMG measures the electrical activity of muscles by inserting a needle through the skin into a muscle to record the electrical activity. The nerve conduction study evaluates nerve function in the arms and legs by measuring the speed with which nerves conduct electrical impulses. A wire is taped on the skin over a nerve to deliver a small electrical shock and another wire taped to the skin records the impulses.
- *Muscle and nerve biopsy*—Diagnoses and characterizes muscle or nerve problems, such as numbness, weakness, muscle cramps, etc. For the muscle biopsy a small piece of muscle tissue about the size of a lima bean is taken from a muscle in the arm or leg for examination under the microscope. For the nerve biopsy a piece of nerve from the ankle is removed. These procedures are done in the operating room under local anesthetic.
- *Ischemic exercise test*—Determines if fatigue after exercise or exercise intolerance and muscle weakness are caused by inability of muscles to effectively use blood sugar—the source of energy for cells. For this test, a small needle is inserted into an arm vein and taped in place, and blood specimens are drawn. Then a tourniquet is placed around the upper arm and the patient squeezes a rubber bulb until he or she either can no longer continue or develops a cramp (this usually occurs within 1 to $1\frac{1}{2}$ minutes). After the exercise, the tourniquet is removed and small amounts of blood are drawn through the same needle at various intervals for up to 10 minutes. The total amount of blood drawn for this test will not exceed 40 ml (less than $1\frac{1}{2}$ ounces).

■ FURTHER STUDY DETAILS:

The objectives of this protocol are: a) screen patients with various neuromuscular disorders and facilitate their entry into appropriate research protocols; b) help resolve puzzling diagnostic neuromuscular problems and train fellows in the evaluation and

(continued on page 13)

European/North American CMT Consortium Meeting To Be Held July 8-10, 2004



The first combined European/North American CMT Consortium Meeting will take place in Antwerp, Belgium from Thursday, July 8th to Saturday, July 10th, 2004. The meeting will have the same format as the previous European and North American CMT Consortium Meetings, with the emphasis on oral presentations and ample time for discussion and interaction. The aim is to exchange new information and, equally important, to start novel collaborations or strengthen existing networks between European and North American research groups.

The program will contain a plenary lecture and introductory lectures covering clinical, diagnostic, and basic research of inherited peripheral neuropathies. An abstract book will be available to participants at the meeting.

The Charcot-Marie-Tooth Association (CMTA) will provide a limited and competitive travel fellowship ($500) to help defray the cost of travel for postdocs who will be presenting at the meeting. A CMTA committee will review and select those abstracts that are eligible for a travel fellowship. ✢

GIFTS WERE MADE

IN MEMORY OF

Kathleen Dannemiller
Debbie Bennett
Anne & Dave Harrell
Gillian Thompson

Nicholas Decoulos
Donna & Jack Altshuler

Irene DeVinney
Evie Safran

Marina Dragonas
Donna & Jack Altshuler

Carol Felt
Robert Fontichiaro

Georgia Cutler
Linda Baron

Bertram Glovsky
Donna & Jack Altshuler

Iris Golmetz
Karen Gabel

Linda Hall
Kathy Carr
Katherine Touchstone

William Long, Jr.
Mr. & Mrs. Robert Long
Richard & Beth Wilkinson

James Luken
Employees of Tower Federal Credit Union

Guy Norton
Mr. & Mrs. Harvey Kerns
William McKee
Daniel & Jane Phillips
Lewis & Linda Sharpes

Richard Rogers
Marilyn Keller
Barbara Schulz

Minna Sand
Peter & Janet Sand Lawner
Michael & Diane Sand

Lamar Stephenson
Helen Mattox

Orville Stocker
Ron & Donna Cornwell
John & Anabel Gallagher
Jean Stocker

Mertice Wallace
Mr. & Mrs. Andrew Brennan

William Wallace
Joan Ebert
Patsy & Jimmy Harris
Bill & Linda Messerly
Toma Norrell
Dr. and Mrs. Daniel Skandera
Charles and Evelyn Smith
Vera Wallace

Dean Weston
Dorothy Helms
Helen Mattox
Mel & Bonnie Rogie
Rebecca Torland

John Whigham
Donna & Jack Altshuler

IN HONOR OF

Zohar Birin
Leon Gelman

Margaret Davis
F.A. & Patricia Davis

Lester Elscott
Associated Wholesale Grocers

Gail and Gaelyn Feeney-Coyle
Bob Feeney

Dorothy Knowles
Herbert Holly

Mr. & Mrs. Hart Wurzburg
Marjorie Grodin
Mary Lawton
Harry & Elsa Levi
Jane Logan
Aimee & Bud Minkin
Hortense Mintzer
Henriette Wolf
Ann & Arnie Wolff

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: ______________________________
____

Excerpts from the CMTA Website Bulletin Board

For those of you who don't have a computer, we are presenting a thread that has recently been posted and which contains many useful ideas that CMT patients have shared. Those of you with Internet access can access the bulletin board at our website (www.charcot-marie-tooth.org) by clicking on "Enter the CMT Chat & Discussion Group." Just reading the posts will give you lots of information, but remember that most of the people posting are not medically trained and you MUST check any advice with your doctor.

I: I was fortunate enough to meet a wonderful surgeon, through my local CMT association, who recommends tendon transfers for CMT patients. I had this operation on both my legs and I could not be happier. She wishes doctors would embrace tendon transfers, rather than bone fusion as a way of managing the symptoms of CMT, as the outcome is much more useful to sufferers. Many orthopaedic surgeons are able to perform tendon transfers, but are not used to doing so for CMT patients. Some doctors think they shouldn't operate on CMT patients because it won't cure or halt the progression of the disease. I could not disagree more strongly. My quality of life has greatly improved by no longer having to wear AFOs. The tendon transfer enables the muscles that push the foot down to be retrained to pull the foot up instead. Has anybody else had a tendon transfer operation on their legs?

S: I haven't, but this sounds really interesting. I may have to research this. I had heard it could be done in the hands.

How bad was your foot drop before you got the surgery? And, did the doctors say if it would last, or will you need AFOs again in the future?

To access the CMT bulletin board, go to the CMTA website and click on "Enter the CMT Chat & Discussion Group" at the upper right-hand side of your window.

C: Surgery for CMT is a very personal decision. I have had tendon transfers done in my left foot. It was a lengthy recovery and it did help for a few years. I will not do my right foot unless there is nothing else to help me. I have found that most surgeries for CMT do help for a while, but we must remember they have only a band-aid effect. I did have a few more years out of my AFOs but the stress on the body from surgery to me is not worth it. I personally will leave any surgery for my last resort. I do fine with my AFOs, get around, and lead a wonderful fulfilling life.

S: I think I'm going to talk this over with my doctor the next time I see him and see what he thinks. It would be nice to be out of AFOs for a few years. I was in them since I was 13, and I'm 21 now. It would be kind of nice to walk down the aisle without them when I get married, but I'm not going to take a huge risk just for that. I'll have a lot to think about now.

C: When I got remarried 4 years ago a big concern was that I wore AFOs. The problem was solved quite easily. I wore a long gown, which, of course, covered the AFOs. I was able to take my white leather shoes and have them dyed to match the color of my gown for when my shoes peeked out from under it. Wearing the AFOs didn't bother me at all because with them I was able to walk down the aisle to the most wonderful man in the world.

S: I still might look into the hand surgery, though. I'm an artist and I'm majoring in art therapy, so I rely on my hands a lot. Right now I'm able to do just about everything I need to, but if the fingers curl up much more, I don't know if I will be able to. If I decide against surgery, or if it won't work out for me, I'm sure I'll find some other way to adapt. We always do. ✳

ASK THE DOCTOR

Dear Doctor,

My 17-year-old son was just diagnosed with Charcot-Marie-Tooth disease (CMT). I've read that "Unlike other neurological disorders, CMT usually isn't life-threatening, and it almost never affects the brain." When it does affect the brain, what are the symptoms? I think my son may be having a problem.

A member of the CMTA's Medical Advisory Board (MAB) replies:

In rare cases, usually sporadic cases with no family history, an MRI of the brain will show abnormalities in white matter or myelin of the brain. This is usually not associated with any clinical symptoms, but since these cases are rare, we have little information about long-term prognosis. CMT2X is associated with mental retardation. This is an X-linked form of CMT. There are also extremely rare cases associated with agenesis of the corpus callosum (a birth defect) that can be associated with developmental delay. You should discuss this with your son's neurologist, who will decide whether or not to order an MRI scan.

Dear Doctor,

Have any positive or negative results been found in the use of glucosamine supplements in individuals with Charcot-Marie-Tooth disease (CMT)? I am considering usage for knee pain and as a preventative for further degeneration of joints.

A member of the CMTA's MAB replies:

Unfortunately, to the best of my knowledge there is no good data supporting the use of glucosamine in treating CMT.

Dear Doctor,

Can patients with CMT develop gastroparesis? My husband is 54 years old with Type 1 with a history of reflux, esophageal strictures, and pyloric stenosis. However, recent EGDs showed no obstruction or narrowing. His symptoms of feeling full and bloated continue. Is gastroparesis a possibility?

A member of the CMTA's MAB replies:

Because neuropathies that affect the vagus nerve could result in gastroparesis, I would seriously consider the possibility that an inherited neuropathy could cause gastroparesis. The demyelinating forms of CMT (type I), however, would not be expected to affect the small axons that are responsible for gastroparesis. Diabetic neuropathy commonly does this, so if the patient is diabetic, I would be inclined to blame the diabetes. Finally, has it been clearly established that the patient really has gastroparesis?

Dear Doctor,

My 50-year-old sister, living in Switzerland, was diagnosed at age 7 with Charcot-Marie-Tooth disease (CMT). She recently had two fractures, one in the heel and one on her tibia, that are not healing. The fractures occurred just from walking on an uneven surface. She was diagnosed with severe osteoporosis just in her legs, with the rest of her body being okay. She is on Fosamax and hormone replacement therapy (HRT). Her fracture sites do not show signs of healing. Do you have any suggestions for therapy?

A member of the CMTA's MAB replies:

This is a difficult question to answer without seeing the patient. I believe she needs to see a physiatrist (rehab physician), a neurologist, or an orthopedist, who are very familiar with Charcot-Marie-Tooth disease (CMT), to best evaluate the patient. Why she is developing fractures so easily, whether this is because of osteoporosis or secondary to her CMT, and how best to treat this requires expertise in caring for CMT.

Dear Doctor,

I am a 50-year-old male with Charcot-Marie-Tooth disease (CMT). Four months ago the middle toe on my left foot became infected. My doctor prescribed antibiotics and a trip to a podiatrist to excise some tissue on the toe. Six weeks later the infection came back and was treated with a different antibiotic and more trips to the podiatrist. Last week the infection came back. Is there a connection between CMT and difficulty with permanently healing this infection? The toe is getting better, but do you think this will be a chronic problem? Is amputation something I may eventually have to worry about?

(continued on page 13)

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

St. Joseph High School Freshman Doesn't Let Disease Slow Him Down

SUSAN OXLEY

Joe Eleniewski is in his first year of high school, an exciting and challenging time for any teen-ager. But Eleniewski also faces the daily challenge of a progressive disease that limits his movement.

Eleniewski was diagnosed with Charcot-Marie-Tooth (CMT), when he was a toddler.

"When I started taking my first steps, I was a lot slower," he said. "I could run when I was younger, but I couldn't walk long distances. I didn't start using a wheelchair until fourth grade."

CMT is a common inherited neurological disorder that affects the nerves that control muscles in the arms and legs. NBA basketball player Todd McCullough was diagnosed with the disease last year.

Joe has an older sister, Tiffany, 18, who does not have CMT. "No one else in my family has it that we know of," he said. "Sometimes the parents don't know they have it until they have a child who is diagnosed with it."

Joe hasn't let CMT slow him down. He earned a 4.0 grade point average his first semester at St. Joseph High School in Kenosha. He also plays wheelchair basketball and wheelchair soccer, and is part of the stage crew for the school's upcoming musical, "Wizard of Oz." He also helps out the junior varsity basketball team by taking statistics at the games.

Joe Eleniewski, fifteen, is a freshman at St. Joseph Highschool in Kenosha, Wisconsin. His favorite quote is from FDR, "All we have to fear is fear itself."

His recent weekly schedule includes daily after-school activities: stage crew, wheelchair soccer practice, mock trial rehearsal, a junior varsity basketball game, and wheelchair basketball practice. On Saturday, he was involved in a mock trial court case in Racine. When asked if he undergoes physical therapy, Eleniewski says, "I don't think I'd have time for it." He said he "plays smart" in his sports activities, resting when he needs to. "I know not to overdo it."

His biggest challenge is transportation, he said, explaining he needs rides from school to after-school activities or from school to home. Snow on the sidewalks in the winter is an added challenge. "My best friend's mom started fund raising for a van that will help us out," he said. "We have a van that we can use until we purchase one." He appreciates the support he finds at St. Joseph High School. "It's like a close family here," he said. "People want to help you out. There's an attitude of generosity."

One of the fund raisers was held in November when St. Joseph High School hosted "Trivia Night" with proceeds benefiting the "Joe on Wheels" fund.

Eleniewski's main interest is computers. He would like to attend Stanford University in California ("There's no snow there") and double-major in computer science and another area of study. He is especially interested in designing computer games. He plans to take the computer classes offered at St. Joseph High School and participate in a program through the University of Wisconsin—Parkside that allows high school students to enroll in advanced computer classes.

At junior high graduation last year, he was pleased to receive the school's "Spirit of St. Joseph" award. "That's not something you earn with your grades; it comes from your personality and what you do outside of school," he said. ✯

(Reprinted from the Catholic Herald *with permission from the author.)*

Truths Children Have Learned

1 No matter how hard you try, you can't baptize cats.

2 When your Mom is mad at your Dad, don't let her brush your hair.

3 You can't trust dogs to watch your food.

4 You can't hide a piece of broccoli in a glass of milk.

5 Puppies still have bad breath even after eating a Tic-Tac.

Truths Adults Have Learned

1 Raising teenagers is like trying to nail Jell-O to a tree.

2 Middle age is when you choose cereal for the fiber, not the toy.

3 If you can remain calm, you don't have all the facts.

4 You're getting old when you stoop to tie your shoes and wonder what else you can do while you're down there.

5 You appreciate the fact that wrinkles don't hurt.

(Editor's Note: These were taken from a publication of insights and inspiration called Good Stuff.*)*

NIH STUDIES

(Continued from page 7)

treatment of Neuromuscular Diseases; and c) provide follow-up to patients who finished their participation in a previous study but are not currently entered in another research protocol. No investigational treatments will be performed on this protocol, but the tissues collected can be used for future research studies.

■ INCLUSION CRITERIA:

Patients may be included in this protocol if: they have an identifiable neuromuscular disorder that may be suitable for a specific current or future protocol; present a diagnostic puzzle in spite of the previously performed diagnostic work-up; or have unusual manifestation of a more usual neuromuscular condition.

Patients should be referred by their doctor, who will obtain our report and will continue patient care.

■ **ENROLLMENT:** 500

■ CONTACT INFORMATION FOR BOTH STUDIES:

National Institute of Neurological Disorders and Stroke (NINDS),
9000 Rockville Pike,
Bethesda, MD 20892;
Recruiting:
Patient Recruitment and Public Liaison Office: 1-800-411-1222
prpl@mail.cc.nih.gov
TTY 1-866-411-1010 ✲

DEAR DOCTOR

(Continued from page 10)

A member of the CMTA's MAB replies:

There certainly is a connection between hereditary neuropathies and foot ulcerations and infections. For instance, Charcot-Marie-Tooth disease (CMT) type 2B and the hereditary sensory neuropathies are closely associated with food ulceration.

Because of the loss of sensation that occurs in CMT and other hereditary neuropathies, microtrauma and more significant traumatic injuries to the foot (stepping on a nail or thumbtack or walking for extended periods with a sharp pebble in the shoe) may go unnoticed. The resulting breakdown of skin and soft tissues in the foot can then lead to infection. If caught early, most of these infections will resolve by removing any offending agents and a course of oral antibiotics. When infections do not respond to such steps, I suggest a referral to a clinic with expertise in wound care. You should also know that foot ulceration can be a major problem in diabetic neuropathy.

Toe and foot amputations are very unlikely in CMT patients. Please don't forget to check your bath or shower water temperature not with your foot, but with a more proximal segment of your body (upper leg or arm, for instance, or have someone else do it for you). And always check your shoes before you put them on! There could be a little rock in there that will create major problems down the road. ✲

LETTERS

Dear CMTA/Pat:

I've been meaning to e-mail you after reading about your horrific accident in the January-February issue of the *CMTA Report.* I'm so sorry that you had to endure all that pain and inconvenience and certainly hope your left arm is progressing to the point where you have full use of it again. Incidentally, the nifty new format with blue panel highlights is excellent and much more contemporary than the previous *CMTA Report.*

We are continuing to revise our website and literature to feature additional products (socks, canes and strutters, heel lifts, braces for sandals, ankle supports, braces for youngsters) and will add a "shopping cart" to our website shortly.

Take care of yourself and watch out for those banana peels!

—Steve Sandler
EZ Gait

Dear CMTA:

We received our latest *CMTA Report* today and we enjoyed it. However, we did miss the "Ask the Dr." column as in the previous issues.

In your "More Helpful Items" section you show a hand-held jar opener. I don't know about other CMT patients, but attempting to hold a jar in one hand, the opener in the other, and successfully opening the jar is something I find impossible. However, I have found one of similar design, but without the handle, that screws under a cabinet that works exceptionally well. They can be found in medical supplies/rehabilitation stores. The ones made of plastic with metal teeth are useless, but the solid metal one is great. Cost is under $20.00 and gives you both hands free to hold the jar and use what "clutch power" we still have left.

—F.S., e-mail

Editor's note: "Ask the Doctor" remains a part of The CMTA Report *and is published periodically. This issue you'll find it on pages 10 and 13.*

Dear CMTA:

I have been diagnosed with CMT, Type 1, and by testing, my neuropathy was diagnosed as severe. I experience a tremendous amount of pain in my feet, legs, and lower back. There is never a time that I don't have some level of pain. I am unable to sit, stand, or lie down for any extended amount of time, and must do each, subsequently throughout the day, including sleeping for approximately 4 hours at a time, sometimes twice a day. Sometimes not.

At times the pain is so bad that I must be admitted to the hospital and treated with morphine to break the cycle. My feet, lower legs up to just below my knees, and hands are numb to the touch, though I do get a sensation that lets me know they're being touched. I experience burning sensations in my feet, knees, and hands. I also experience a great amount of fatigue. I have braces and walk with a cane. I am no longer able to work, and barely able to leave the house.

I am being treated with the following drugs: Trileptal 1200 mg twice a day, Neurontin 400 mg 3 times a day, and since my last hospital visit, hydromorphone 2 mg every 4 hours (previously, I was prescribed hydrocodone 5/500 every 6 hours as needed). The Trileptal and Neurontin seem to do something, but not even close to enough. I am basing this on the fact that, if I am a few hours late on taking these drugs, I feel much worse. The hydrocodone very slightly took the edge off the pain. The hydromorphone is working better, but there is still an appreciable amount of pain.

One of the largest problems I am facing is actually my doctors. Over and over again, I am being told that pain and CMT are not associated with one another! Furthermore, when I discuss pain medication with my doctor and neurologist, they are very reluctant to prescribe them, pointing out that these drugs may be addicting! It is at these times that I am told that there is no, or at best, slight pain, with CMT. I have been tested by two different neurologists and have undergone genetic testing by a third party: all confirmed CMT1. I have been tested for any possible acquired neuropathies, and none have been found. I am telling you, and everyone else, that there are cases of severe pain associated with this disorder/disease. I am living and breathing proof!

The first time I saw my doctor, my symptoms were pain

WRITE TO US!

Pat Dreibelbis, Editor
The CMTA Report
CMTA
2700 Chestnut Pkwy.
Chester, PA 19013
or
CMTAssoc@aol.com

The CMTA reserves the right to edit letters for space.

and fatigue. I was tested and told I had CMT1, not the other way around. Quite frankly, I had not even heard of CMT at that time. Why then, would any doctor question the very reason of one's original visit? Pain medications must be used for management of pain which absolutely never goes away; therefore, addiction is not an issue, because you can never stop taking them anyway. Living with pain at literally every single moment of one's life is a nightmare. Anyone who is experiencing what I am will tell you that there is absolutely no joy in taking these medications. Abuse of these medications is not the issue. Relief of this nightmare is!

Furthermore, I have very easily found many case studies, medical articles, and papers from top neurologists and medical schools from around the world on the Internet that confirm this issue (all I did was type "Charcot Marie Tooth" into the internet search engine on my toolbar). I have printed them out and intend on taking them to my doctor and neurologist. I have also noticed that these problems I am facing with my doctors are not uncommon! I have read many articles from other people who say they have this exact issue with their doctors. Why, if this information is readily available, is it so scarce in the medical community? It is my opinion that doctors should be listening to what their patients are telling them is wrong, not telling their patients what is not wrong. The medical community needs this information desperately..

—K.M., e-mail

Dear CMTA,

I teach children identified with learning disabilities. In an effort to motivate them with interesting activities while providing them with opportunities to reinforce their math, reading, and writing skills, I developed a unit in which my fourth grade students made and sold mason jars filled with cookie mixes for the holiday season. They solicited orders from family members and teaching staff and then created the gifts with their own hands.

The students and I discussed the true meaning of the holiday season—the spirit of giving to others. The students then were each given the opportunity to select a charity of his/her own choice to send a portion of the project's earnings. Your charity was one of those chosen by the fourth graders.

I hope the enclosed donation is a help for your organization's work. It was raised by a very special group of children who were able to not only practice their math, reading and writing skills, but also learn the important lesson of helping others.

—D.M.
Learning Support Teacher
Amosland Elementary School
Morton, PA

Dear Pat/CMTA,

Your great article on stubbornness and disability prompted me to write to you.

I'm a 70-year-old female, diagnosed in 1993, after symptoms that began in the late 1980s. I have noticeable muscle wasting in my legs, feet, arms, and hands. I use a cane now, but my doctor wants me to use a walker. Vanity and stubbornness stop me from using it.

I was extremely impressed with how you improvised, in spite of all your problems. I'd like to share with you how I keep active, despite my walking difficulties and very poor balance.

When the weather is nice, I can still ride my bike (a small one). Getting on and off can be a problem, however.

Every year I book a cruise with my friends. This past February, we cruised the Panama Canal. The side tours which I did were sailing, with lunch on the beach, Catamaran, and snorkeling off the boat (the flippers were my biggest problem, so I sat on my butt and scooted over to the steps and went down into the water).

Wow! What a feeling of accomplishment. I also went horseback riding on the beach—I just needed a boost up. I had a ball.

My motto is "keep a good and positive outlook—keep trying everything and don't let people tell you 'you're not able to do that.' "

—J. P. Cape May, NJ

Dear CMTA:

I would like to share the shoes that I wear with my AFO. They are the New Balance Walking Shoes. I purchased them at Mason's Shoe Comfort Footwear, 1251 1st Avenue, Chippewa Falls, WI 54774. Phone No. (1-800-428-2000) The price is $79.95. Sizes run from AA, B, D, EE through EEEE (6 to 13). They can be found in most athletic shoe stores and many department stores.

—G.L., e-mail

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

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.

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.

THE CMTA *Report*

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