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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

Welcome to the new CMTA website!

Yes, at long last here we are, albeit in what's known as the "beta" version. That means things work more or less as they are expected to work, but there are still a few bugs and glitches to be worked out. We'll be identifying and correcting them over the next several weeks, and we apologize for any inconvenience.

When we started this project late last year, our goal was to provide you with more up-to-date information than ever before in an easily accessible format. Let's take a quick tour of the new features, starting with the column of links on the left side of the homepage.

■ **About CMT:** From the Overview to the page on CMT and Pain, this section has much more information on all aspects of CMT.

■ **Ask the Experts:** We've added many more questions and answers and grouped them by topic. Once you select a topic, clicking on a question brings you right to the answer, or you can scroll down the entire page and read all the questions and answers under that topic. (You can also find specific questions using the site search box.)

If you have a specific question you'd like to ask a member of our Medical Advisory Board, click on the link and email it to us at asktheexpert@charcot-marie-tooth.org. We'll forward it to the appropriate doctor and reply directly to you.

■ **Medical Alert:** This page displays our standard medical alert list, but members who are logged in can also view an expanded version of the list. (More on logging in later.)

■ **Resources:** Got a comfortable chair? You can spend hours exploring the resources compiled here. Start by entering your state and then select any of the categories. Physician lists and support groups will be displayed by state only, but the other categories will display results for both the state you selected and for national or international agencies and organizations.

We'd also like to mention that you can make a valuable contribution to the CMTA and to everyone who uses these resources. This information is constantly changing, especially the physician lists, and there are also areas for which we have very little information. If you know physicians who are knowledgeable about CMT but who are not on the list, or if any of the other information is incorrect, please email us at webmaster@charcot-marie-tooth.org.

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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

INSIDE:

The CMTA Visits Capitol Hill

The CMTA continued its work with the federal government as the Board of Directors met in Washington, DC, on April 26th and 27th. Building on the success of 2004, when strong language on CMT was included in a key Congressional Committee Report, the CMTA Board is working with the National Institutes of Health (NIH) and members of Congress and the Senate to underline the importance of federal support for CMT research.

On April 26th, Dr. Michael Shy, Chairman of the CMTA's Medical Advisory Board, President Patrick Torchia, and Executive Director Charles F. Hagins visited the National Institute of Neurological Disorders and Stroke (NINDS) of the NIH to discuss recent advances in CMT research and the February 2005 NIH report on CMT research.

J. Rodman Steele, Jason Steinbaum, Robert Kleinman, Patrick Torchia, Dr. Michael Shy, Phyllis Sanders, Charles F. Hagins, and Steve O'Donnell met with government representatives to push for more CMT funding.

North American CMT Consortium Meets in Canada

BY CHARLES F. HAGINS

For three days, the air was alive with talk of genetics and pure science. Meeting in London, Ontario, Canada, at the Spenser Conference Centre, the Symposium of the North American CMT Consortium took place from May 19 to 22. Over forty of the world's leading CMT researchers presented their scientific findings to the group for critique, suggestions, and encouragement.

The invited guest lecturer was Dr. Lawrence Wrabetz from the San Raffaele Scientific Institute in Milan, Italy. He began the scientific presentations with the Ann Lee Beyer Plenary Address entitled, "Gain of abnormal function and phenotypic diversity in MPZ-related neuropathies."

During the morning coffee break, Dr. Wrabetz spoke with me and said, "This is an amazing event! I've already received two suggestions regarding my scientific findings that I never considered!" Clearly, this is the

Executive Director Charles F. Hagins presented Dr. Lawrence Wrabetz with a plaque for his delivery of the Ann Lee Beyer Plenary Address.

At the meeting with the NINDS director and high-level scientists, Dr. Shy offered extensive detail about the importance and promise of CMT research. The NINDS leadership asked many good questions and encouraged the CMTA group to continue along the path they are pursuing.

The next day, the board members traveled to Capitol Hill where they met with the staff of several Senators and Members of Congress. In fact, the Board's meeting was attended by the legislative director of Representative Curt Weldon (R-PA) who represents the CMTA's office in Chester, PA. The CMTA greatly appreciates Rep. Weldon's support of efforts to increase CMT research dollars. He and Rep. Eliot Engel (D-NY) are leading the charge in the House of Representatives to increase CMT research money. ✲

object of such meetings—to allow researchers to assemble and discuss what they are doing in the presence of others with similar knowledge, rather than working in isolation.

Collaborative efforts have arisen from these consortiums in the past and, after two full days of scientific presentations, I'm sure that, once again, the molecular and genetic science behind CMT has been advanced significantly.

This event was organized and chaired by Dr. Michael Shy, Chairman of the CMTA's Medical Advisory Board. ✲

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**	
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			

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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.

NEW WEBSITE

(Continued from page 1)

■ **Daily Living Aids:** We're always amazed by the ways people have found to make everyday tasks easier, but we're equally amazed how many people are unaware of the Aids for Daily Living (ADLs) that can make those tasks easier and safer. We have partnered with ActiveForever to bring you a selection of ADLs that people with CMT find most helpful. Whenever you "click through" an item on this page and purchase it or any other product from ActiveForever, they will donate 10 percent of the purchase price to the CMTA, so you'll be helping us, too.

■ **Discussion Forums:** The creaky old Ultimate Bulletin Board is no more. We've done away with its hodgepodge forums and replaced them with five general forums organized around specific topics and a sixth general forum for "personal thoughts."

Members will be able to access two additional forums, one called CMTA ParentsNet and one called CMTA KidsNet. While we work to prepare information for schools, raise funds, and organize events, the ParentsNet will be a place to share information and ideas. If you're a parent, you're welcome to join us!

The KidsNet also has a special purpose. We've seen how important it is for kids to be able to connect with their peers and talk about how CMT impacts their lives, and we wanted a safe and secure place for them to exchange messages.

■ **Research:** The CMTA is committed to funding research and finding a cure. On this page you can follow our funding history year by year. You'll see we started small, but we've now funded almost $2 million in research and this year we're increasing the number and size of research grants awarded, so watch this space!

■ **CMT Database:** We also help fund the CMT North American Database begun by Wayne State University and maintained at Indiana University. It's an important tool for scientists and researchers, and you can read more about it here, as well as find out how to enroll.

■ **Parents and Kids:** We're just getting started on these pages. The articles and stories here are contributed by people with CMT. We hope they'll become an important resource and provide people with inspiration and insight.

■ **Archives:** The first issue of *The NFPMA Report* came out in 1987—it wouldn't become *The CMTA Report* until 1990—and members can download and read PDFs of every issue. Select a year, and the tables of contents will be displayed, or, enter a keyword and search. Either way, you'll need that comfortable chair again!

We're also creating an archive of headline articles from the homepage, so members will be able to view them as well.

■ **Events:** Oops. This page needs a little more work. It will eventually be two pages, one where you can read about upcoming events and one where you can read what happened at past events.

■ **Email List:** The less we have to spend on postage and printing, the more we can spend on other operations and research. Email communication is the most efficient and cost-effective way to get information to you. Sign up today!

How's that chair? Still comfy? Good, let's go to the menu bar of links at the top of the homepage.

■ **About the CMTA:** We ask for your money and your support, so we'll tell you what our mission is and how we'll spend your money. You can meet our Board of Directors and see why they have both the professional competence to govern the organization and the personal commitment to find a cure. Our Medical Advisory Board is here, too, and you can read about our history.

■ **Join the CMTA:** Okay, this isn't totally new, but there are some important details for both prospective and current members. One is that when you join or renew your membership, you need to indicate how you want

RESPIRATORY PHYSICIANS FOR PEOPLE WITH NEUROMUSCULAR PROBLEMS

Physicians with an interest in the respiratory care of people with muscular dystrophy and neuromuscular weakness have agreed to have their contact information made available online for The Parent Project Muscular Dystrophy site *(www.parentprojectmd.org)*. Most of these physicians require a referral from a primary care doctor. The site is maintained by Jonathan D. Finder, MD, Children's Hospital of Pittsburgh. *(www.pitt.edu/~finder/respdocMD.html)*

to receive *The CMTA Report*. If you choose the "PDF via Email" option, we'll email you a link that will enable you to read the newsletter as soon as it's compiled. Or you can specify "Print" and we'll mail you a copy. If you choose "Print and PDF," you'll get both, but your dues will be $45 instead of $40.

Second, you must enter a valid email address if you want to receive the PDF version of the newsletter or access the members-only areas of the website.

Finally, if you choose to make an additional donation, you may choose to have it used for operations or for research.

■ **How to Contribute:** Membership, for those able to pay the dues, is a vital means of support. It's what keeps the lights on and the phones working, and it allows us to provide information and services to the many people with CMT who are on disability or struggling financially.

The fight against CMT requires much more, however, and many of you support us generously through our annual and research appeals. Email and our secure online donation form will help make that process more cost-effective, too.

If you are making a donation in honor or in memory of someone, please enter the person's name and indicate to whom the acknowledgment should be sent.

If you make purchases online, you can support us by registering with *iGive.com* and identifying the CMTA as your cause. Soon, you will also be able to shop on line directly from our affiliates page.

Finally, we encourage you to consider other ways to support the CMTA, such as donations of stock or testamentary bequests.

■ **Search:** Enter a keyword or phrase and click "Go" to search the site. Cool.

■ **Member Log-in/Log-out:** When you visit the site, you'll want to take a moment and log in so you have access to all areas, but first you need a username and password.

If you're new and join online, you will be asked to enter a valid email address, a username, and a password. You will be registered automatically and can log in right away. (We require email so the automated system can send you a new password if you forget yours.)

If you're a current member, you'll have to go to the forum and register. Once you've done that, send an email to username@charcot-marie-tooth.org and tell us your username. You will have access the next business day.

■ **About the Flash:** Last but not least, we'd like to give a generous round of thanks to the people who appear in the Flash photos. They are truly the faces of CMT. ✲

GIFTS WERE MADE TO THE CMTA

IN HONOR OF

Yohan Bouchard
North End Express Deli

Joe Gelman
Leon Gelman

Zack Shapiro
Deborah & Oliver Shapiro

IN MEMORY OF

Richard Aishton
Florence Braverman
Karl Falter
Thomas & Margaret Lillard
Joan & Harvey Meyers
Janet Raciti
Marlene & Henry Wasserstrom

Harry Berkovitz
Sarah Berkovitz

Ann Lee Beyer
Caroline Durr

Betty Chow
Edie & Rex

Kathleen Dannemiller
Robert Stephens

Hubert Firnhaber
Helen Firnhaber

Walter Flowers
Jeff & Lisa Matheny

Henry Gemeny
Marshall Street School Staff

Jeff Gerry
Fred & Amy Miller

Mrs. Horsmann
Geralyn Carroll

Ruth Linker
Patricia Beaumont
Larry & Judith Brenner
John & Karen Campbell
Anne & Thomas DeFranza
Gregg Linker
Kristen Schaefer
Michele & Eugene Sullivan

Mario Palombo
Maria Cipriano

Donna Pennington
Kathy Car
Katherine Touchstone

Roger VanderBoom
Sandra Lee Barton
Rita Clauss
Bob & Kayleen Lee
Ruth Overley
Katherine Reichstein

Thoughts on CMT and Parenting

BY KIM FORD AND ROB WERLING

We would like to share with the CMT community some of our experiences, thoughts, fears, and strategies for living with a child with CMT. We have included a little history about our "discovery" of CMT, followed by a description of the practical impact it has had on our lives, some general thoughts and feelings about the journey with this neuropathy, and finally a few small practical points specific to school-age children. When all is said and done, our message is encouragement to make the most of life's vicissitudes: the good, the bad, and the indifferent.

FEAR OF THE UNKNOWN

Six years ago, when Keenon was 5, his pediatrician noticed something we had been vaguely aware of, but not particularly worried about. He had always been perfectly healthy, hitting landmarks on time and impressing everyone with his articulateness and sparkle as a toddler, but the pediatrician in Seattle said in no uncertain terms that Keenon's foot-drop gait was not normal and needed to be evaluated.

That assessment set us on the journey we are still navigating today, without a map or a compass or an owner's manual. With the 5-year-old Keenon, we spent hours, days—months—with neurologists, pediatric neurologists, neuromuscular physiologists, and geneticists. After ruling out spinal cord tumors and metabolic deficiencies and goodness-knows-what-else, the unanimous opinion from the best minds in the business was that Keenon "probably has some type of CMT."

Keenon Ford was diagnosed with CMT at age five.

"What should we expect?" we asked. "What does that mean?" Whether they told us directly that they didn't know the answers or avoided the questions, it became clear to us that we were dealing with what one neurologist called "Keenon's disease," that this was going to be a unique journey, and that we would see what to expect when it happened.

As parents, we had nightmares and tears of dread, thinking about how our little boy, who could still struggle awkwardly to keep up with his peers on the soccer field, would one day be wheelchair-bound. Or that this kid, who can make intricate 3-dimensional structures out of paper and draw life-like portraits, might one day have non-functioning hands. Over the years, as we have lived with the fears and compared them with what is actually evolving, they have become less painful, but they will never go away completely.

IMPACT ON KEENON'S LIFE

For Keenon, life just goes on. In contrast to our experience, fear of future problems is not an obvious part of the impact of CMT on his life. The practical effects on his life can be divided into three categories: (1) adjustments in fine motor activities, (2) large motor activity limitations, and (3) maintenance.

■ **Fine motor.** Keenon's hand strength is less than normal for his age. Examples of the practical impact of this include that his fingers shake when he works for too long on a paper model or handwriting. So far, he has not had trouble with standardized tests, etc. Occasionally, his hands cramp painfully, making some activities essentially impossible. Recent examples include riding an electric scooter with stiff brake and accelerator levers, and participating in a junior SCUBA certification course. Part of that course was in a Minnesota lake in October, which meant the water was very cold. Certain tasks in diving require manual dexterity, which simply did not exist for Keenon under those conditions. (We spent a few anxious moments, but once we had warm water to work in, everything was fine.)

■ **Large motor.** More than the effects on his hands, Keenon's reduced mobility on his feet influences his life. With the help of AFOs (we call them "splints"), he can walk short distances—around the house or classroom—just like anyone. With longer distances, though, his pace becomes too slow to match that of his peers and family members. He also tires quickly at swimming and biking, even though these activities don't directly involve the muscles most affected by CMT. He has not participated in organized sports (like soccer, which he really enjoyed) since age 6 or so.

There are times when these limitations make our whole world seem bleak. For example, when the casts (for stretching his very tight heel cords) were removed from both his legs, he could not walk at all for a few days, and had to crawl to the bathroom. But most of the time CMT is more like that irritating stain you can't remove from the carpet—something that just is.

Socially, Keenon seems to fit in at school, despite much decreased participation in gym and recess activities. (He sometimes "meditates" while the other kids are playing football or whatever.) He is physically different from his peers, but in the classroom his differences are subtle enough that his personality is more important in shaping relationships.

■ **Maintenance.** The biggest practical daily impact of CMT on Keenon's life is management of AFOs, physical therapy, and annual month-long casts which help to loosen his very tight heel cords. Since he continues to grow like any 11-year old, the splints get too small; they break, or rivets pop off (to be replaced by duct tape), or they cause blisters or painful infected "hangnails." The management of his stretching and physical activity has become a big part of our CMT experience because Keenon does not yet fully grasp the long-term benefits of a vigilant physical therapy program.

Gillette Children's Hospital in Saint Paul, MN, has been our CMT outpost for the past two years. The care and assistance we have received at Gillette Children's Hospital have been outstanding. Gillette specializes in pediatric orthopedics. It is run like a small company, where the different subspecialists all confer with one another and become a working team to help each child be the most active he can. They work hard to customize Keenon's AFOs to give him excellent support without diminishing his quality of life and mobility. Their attention to his needs, willingness to be creative and "think out of the box," and commitment to make him satisfied are fantastic.

OUR SHARING AND COPING TECHNIQUES

1. Present (at the beginning of each school year) a brief explanation of why Keenon wears AFOs and hold a question-and-answer session on CMT.
2. Keep school personnel very informed. We give teachers a packet of general information on CMT and how Keenon is affected. (Includes doctor contact info.)
3. Establish a special physical fitness program as an alternative to strenuous physical education classes.
4. Establish an alternative indoor recess activity when Keenon is casted and immobile—working in the computer lab, working in the library, doing a special math project, etc.
5. Work with the same people at Gillette Children's Hospital, and keep actively involved in splint options, stem cell research, etc.
6. Establish a physical therapy/fitness plan with a YMCA trainer to build Keenon's confidence about his body.
7. Swim regularly…stretch daily…yoga…explore the benefits of pilates and yoga…
8. Be postive…and laugh…the glass is half full!

STRATEGIES

Keenon is physically challenged, but he is blessed with an active and inquisitive mind, and he excels at so many non-physical things that his world is wonderful. In fact, we wonder whether he would have discovered some of his interests if he were more active. With the advent of the electronic and technical age, it is a great time to be logging hours on a computer, 'Googling' information, creating stories and movies. Opportunities abound to discover and excel in all different fields: the arts, technology, cuisine, building, music.

Keenon's energy is devoted to reading, computers, chess,

(continued on page 12)

The Kindness of Strangers

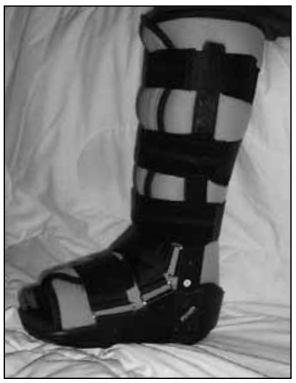
BY PAT DREIBELBIS

When last I wrote a personal story for this newsletter, I was retelling the story of my fall and broken elbow, the subsequent insertion of a rod and steel pins, and both the helplessness of being one-armed and the sense of sheer (and mindless) determination with which I tried to do tasks related to getting ready for Thanksgiving and Christmas.

This personal story also revolves around disaster (I'm apparently prone to such things on a biannual basis) and also involves the frustration of being handicapped (albeit temporarily) and unable to do the things I normally could. I had been having sharp and unexplained pains in my left foot around the arch and the ankle. I attributed it to nothing more than an aspect of my diabetes, or just standing too much in stores and the kitchen. After four weeks of basically pretending that nothing serious could actually be wrong, I gave up and went to a podiatrist. After several sets of x-rays, it was determined that I had a broken bone (the navicular) and that I would need to be immobilized. This was not the "right" answer because I was facing a very busy spring with my daughter graduating from medical school and my son, one day later, from college. I really had no time to be in a cast, even if it was the walking variety.

Undaunted by my whining or my story of how this cast was going to ruin my life, the podiatrist sent me to Hanger Orthopaedics to be fitted for the contraption in which I was doomed to spend at least six weeks.

I have to say that my first few hours in the cast had me believing that I would probably fall and break something else if I actually stayed in the cast because it had a rocker bottom and it was very difficult to adjust to. Two weeks later, I think I finally have the gist of walking, but I really can't navigate on uneven ground or go up or down stairs with any confidence or speed.



***CMTA Report* editor Pat Dreibelbis was rendered incapacitated by this "contraption."**

I have found, however, that having an ungainly and unattractive gait and appliance on my leg has produced all varieties of kindness. In a restaurant the other night, with waiting lines, a man got up from his chair and offered it to me. Men and women hold doors open for me. Even my coworkers jump up from their chairs to bring me faxes and mail so that I won't have to walk around unnecessarily. I've even managed to get a friend to do my grocery shopping so I don't have to stand or walk for long periods of time.

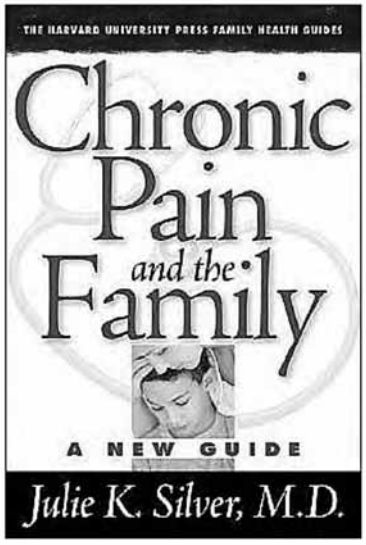
Good for me….but there must be some reason for this article other than a simple retelling of yet another disaster in my life. What I've come to realize is that CMT, which for most patients is a fairly invisible disorder, garners people very little "kindness" from strangers. Unlike my obvious disability, CMT quietly makes it difficult for someone to stand in long waiting lines. It makes walking on uneven ground a real challenge and I'm sure that grocery shopping for someone with CMT isn't much fun. Since fatigue is such an issue, all the tasks that are formidable for me right now are always formidable for someone with CMT. But, I'm getting lots of sympathy and empathy, things which probably aren't readily available to people with CMT. Do you want sympathy or empathy? I'm not sure. I am sure you deserve it more than I do.

When I broke my elbow and struggled with one-handedness, I thought maybe I was being given a chance to understand at least a little more what it's like to have a disability. Now that I've broken my foot, I think I'm getting even closer to understanding and being able to empathize with patients who have CMT. I don't think I need any more lessons…at least, I sure hope I don't. ✲

Books Worth a Look

CHRONIC PAIN AND THE FAMILY, A NEW GUIDE

Julie K. Silver, M.D.



Chronic pain is the leading cause of disability in the United States, affecting as many as 48 million people in this country alone. It can demoralize and depress both patient and family, especially when there is no effective pain control and no hope for relief. Improperly managed, chronic pain can lead to substance abuse (usually pain killers) and to acute psychological and emotional distress. Pain begets stress and stress begets pain in a wretched downward spiral.

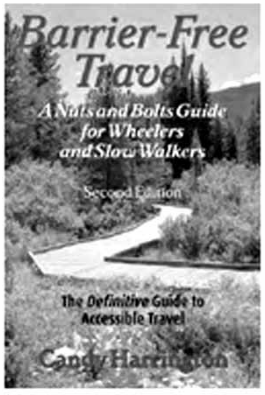
Silver reviews the causes and characteristics of chronic pain and explores its impact on individual family relationships and on the extended family, covering such issues as employment, parenting, childbearing and inheritance, and emotional health. Silver treats aspects of chronic pain not covered in a typical office visit: how men and women differ in their experience of chronic pain, the effect of chronic pain on a toddler's behavior or an older child's performance in school, the risks of dependence and on addiction to pain medications, and practical ways for relatives beyond the immediate family circle to offer help and support to the person in pain.

Silver is the Medical Director of International Rehabilitation Center for Polio at Spaulding-Framingham Outpatient Center, and Assistant Professor of Physical Medicine and Rehabilitation at Harvard University.

■ *Available from Harvard University Press, www.hup.harvard.edu/catalog, ISBN 0-674-01-666-1, $14.00 paperback*

BARRIER-FREE TRAVEL: A NUTS AND BOLTS GUIDE FOR WHEELERS AND SLOW WALKERS

2nd Edition, Candy Harrington



Authored by the editor of the leading travel magazine for people with disabilities, Emerging Horizons, this second edition of Barrier-Free Travel is the definitive guide to accessible travel for those who use a wheelchair, walker, or cane or have any physical ailment that may slow down their gait. It is a well-researched resource that contains detailed information about the logistics of planning accessible travel by plane, train, bus, or ship. It contains resources, travel tips, and updated information about accessible travel options.

Chapters include information on how to find and book an accessible room, cruises, ground transportation, overseas travel, advocacy, disability law and rights, air travel, protecting your wheelchair when you travel, how to find and work with a travel agent, and resources from around the world. This new edition includes a chapter on children's travel issues and a much expanded section on cruises. Expanded airport security procedures have changed the way we travel and thus the book offers new information about these security procedures and how they apply to wheelchair-users and slow walkers.

Barrier-Free Travel contains essential information—not just common-sense tips. Because of the chaos and uncertainty involved in travel, people who require access accommodations need thoughtful, reliable information. This book gives readers the tools and resources to prepare for their journeys and is essential reading for every traveler with mobility limitations.

■ *Available from Demos Medical Publishing, www.demosmedpub.com or call 1-800-532-8663, $16.95 soft cover* ✲

Three Research Grants Awarded by CMTA

The first three-year, $100,000/year grant awarded by the Charcot-Marie-Tooth Association has been given to Dr. James Lupski, a member of our Medical Advisory Board and a Professor of Molecular and Human Genetics at the Baylor College of Medicine in Houston, Texas.

Dr. Lupski's grant is entitled "Molecular therapy and management of CMT." His objective is to understand the molecular genetic basis of Charcot-Marie-Tooth disease and related peripheral neuropathies and, based on that understanding, develop new therapeutic approaches. He will study the efficacy of small inhibitory RNAs to cancel PMP22 over-expression seen in patients with CMT1A, the type affecting 70% to 90% of all CMT patients.

The other two grants awarded for this year are $35,000 and have been given to Dr. Yann Saint-Georges and Dr. Anthony Antonellis.

Dr. Saint-Georges is from the University of Utah, the Department of Biochemistry. His grant is entitled, "Functional analysis of MFN2 mutations implicated in Charcot-Marie-Tooth type 2 (CMT2A) neuropathy." Dr. Saint-Georges will study mutations in the human mitofusin 2 gene (Mfn2) which is linked to CMT2A. Mfn2 encodes a protein required for fusion of mitochondria, a cell compartment necessary for cell viability. Proteins sharing the same function with human Mfn2 can be found in many organisms like yeast, worms, flies, and mice. Much of what is now known about Mfn2 function is derived from studies of

CHALLENGE GRANT PROGRESS

The Challenge issued by Board Member Gary Gasper has already generated over almost $70,000 in matching funds. Please join the challenge and help us make, and exceed, our goal. Watch the mail for the research fundraising letter, which encourages each of you to work on making the $100,000 from Gary into $200,000 or more!

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:_____

_____

budding yeast. The experiments will help to define the molecular role of Mfn2 during mitochondrial fusion and will provide new information regarding the defects of Mfn2 that are associated with CMT.

The final recipient, Dr. Anthony Antonellis, is a postdoctoral fellow at the National Human Genome Institute in Bethesda, MD. His grant is entitled, "Functional analysis of GARS mutations identified in patients with CMT2D and dSMA-V." Charcot-Marie-Tooth disease type 2D and distal spinal muscular atrophy type V are characterized by muscle weakness in the arms and legs, which is more severe in the hands. Previous efforts showed that both diseases are caused by mutations in the glycl-tRNA synthetase gene (GARS). The enzyme encoded by GARS is responsible for a major step in protein synthesis, which is essential for living cells and organisms.

Defects in this protein cause a neurological disease specific to the hands and feet, with few other detrimental effects. The study will determine if GARS mutations affect protein synthesis and may reveal a special requirement for this process in CMT-affected cells.

The awarding of these three grants, totaling $170,000 for this year, makes your giving to the Challenge Grant Research Fund even more important. If you haven't done so yet, please return your research envelope with a generous gift. ✲

A WORD ABOUT CHARITIES

(From NORD ON-Line bulletin)

American charities are having a tough time, not because a slow economy has diminished donations from the American public, but because epic tragedies have drawn the public's attention and donations away from non-profit agencies that have traditionally benefited from the public's generosity.

The September 11 calamity raised record-high donations four years ago as Americans rushed to help surviving families. As a result, in 2001 many traditional charities were unable to meet their budgets. The following years indicated a slow economic recovery due to the recession, which meant that families didn't have extra money to donate to charities. In 2004, traditional charities had to compete with political fundraising campaigns. Then at the end of 2004, the tsunamis occurred and again Americans showed their heartfelt compassion for the millions of people who suffered in that epic human tragedy.

Unfortunately, many American charities that often rely on end-of-year gifts from generous donors struggled at the end of 2004. Donations were not good for all except the international aid organizations.

The Chronicle of Philanthropy says, "The bleak outlook for 2005 comes after a year in which many charities struggled to raise enough money to keep pace with inflation, which was 3.3 percent in 2004". Many non-profits that have relied on government funding have suffered deep and painful cuts in recent years, except for "faith-based" organizations that are newly eligible for federal funding. But, as state and federal governments decrease their support for social services, charities are expected to pick up the slack even though they have similar budgets to tackle bigger jobs.

We suspect two things have to happen to improve the outlook for U.S. charities: First, we must get the attention of the American public and show them how valuable our services are (especially for medically disenfranchised people with unusual diseases and unusual problems). Secondly, we have to hope that there will be a true economic turnaround quickly. People who are worried that they may lose their job, and those who have no job, are unwilling or unable to donate to charity. When the economy is good, charities prosper, especially at the end of the year when people think about tax deductions.

Ultimately, the major catastrophes that have drawn the public's attention and donations in the last few years have also taught an important lesson for all of us: The American public can be passionately humane and generous when it recognizes a worthy cause. Getting to the top of the public's list of worthy causes is the challenge! ✲

SUPPORT GROUP NEWS

New York – Horseheads

Our latest support group meeting was held on May 11. We had a total of 9 people in attendance. Our speaker for the evening had to cancel at the last minute, so we sat and discussed our everyday problems and the solutions we had found for some of them. One thing which came out was the frustration we all have due to the lack of help with our symptoms by our "local" physicians. Two or three of our group are now living check to check, SSI, or disability, etc., which makes things so much more difficult.

One of our members, Bill Stanton, 83 years old, recently bowled a 505 series in the Chemung County Senior Games. He received a medal for his efforts. We are so proud of Bill and his wife, Donna, who accompanies him to our meetings and many other events, in spite of severe osteoporosis.

Pennsylvania – Johnstown

The Johnstown, PA CMTA Support Group held its bimonthly meeting April 28 at The John P. Murtha Neuroscience and Pain Institute. John Letizia from Laurel Medical discussed aids and helpful devices to make life easier for people with CMT. John is all too familiar with CMT. His uncle is Don McLaughlin, the patriarch of our CMT group.

Ten years ago, Don singlehandedly assembled 30 members of his family in Johnstown one Sunday afternoon to allow Dr. Jeffery Vance and his team from Duke University to draw their blood for genetic analysis. The family's genes, and genes from six other large families, including families from Russia, Italy, Turkey, and Japan, were used to determine the genetic cause of CMT2A. In the April 2004 issue of *Nature Genetics*, an international group led by Stephan Zuchner, MD, and Jeffery Vance, MD, announced, "Our results indicate that mitofusin 2 (MFN2) is a major gene underlying CMT type 2A, and probably one of the major genes that cause all hereditary forms of the axonal neuropathy, CMT type 2." As a result of this discovery, Athena Diagnostics now offers genetic testing for CMT2A and researchers are now able to search for therapies to cure the disease.

Don McLaughlin, seated with grandson, Kellan, faithfully attends the Johnstown Support Group Meetings.

CMT has dealt Don a particularly nasty hand. He has been wheelchair-bound for years and his breathing muscles are so weak he is forced to use a BiPAP at night. Don, unfortunately, is presently in Conemaugh Hospital following a tracheotomy and is trying to wean himself from a respirator. He is one tough man—a former football star and tank commander. We wish him our best. He is in our prayers and we sincerely thank him for all he has done for CMT. ✲

KEENON

(Continued from page 7)

cello, graphic animation, acting, writing, drawing, and math. As CMT ebbs and flows through our life, we are supportive and emotional, but we will not let it control our destiny. Our goal is to help Keenon cultivate a PASSION and inner drive to pursue one of these things as an adult.

No matter what Keenon's physical condition, we struggle like all parents to make sure he has a solid foundation of morals, acts responsibly, resolves conflict with words and reaches out to those who feel left out. We strive to focus on who Keenon is, not how he moves about the world. It is not about what we can do for him but what he can learn to do for himself. How can he become independent and self-satisfied? This challenge is made more difficult with the added factor that Keenon has CMT, but maybe that makes us better parents, too. ✲

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—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

Florida—Tampa Bay Area
Place: St. Anthony's Hospital, St. Petersburg, FL
Meeting: Quarterly, 2nd Saturday, 10:30 AM
Contact: Lori Rath, 727-784-7455
Email: rathouse1@msn.com

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

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

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 every other month, 1-3 PM
Contact: Dr. David Younger, 212-535-4314, Fax 212-535-6392
Website: www.cmtnyc.org
Email: bwine@acm.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: Bimonthly, Jan, March, May, Sept, and Nov; 3rd Saturday
Contacts: Beverly Wurzel, 845-783-2815
Eileen Spell, 201-447-2183
Email: cranomat@frontiernet.net espell@optonline.net

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: Wills Restaurant 405 Wagner Ave, Greenville
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: Darlene Weston, 503-245-8444
Email: blzerbabe@aol.com

Pennsylvania—Johnstown Area
Place: Crichton Center for Advanced Rehabilitation
Meeting: Bimonthly
Contact: J. D. Griffith, 814-539-2341
Jeana Sweeney, 814-262-8467
Email: jdgriffith@atlanticbb.net, cjsweeney@ussco.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, 732-977-9983
Email: astarryoung@yahoo.com

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

ASK THE DOCTOR

Dear Doctor:

I am currently taking 75 mg of Effexor, an antidepressant described to me as a selective serotonin-reuptake inhibitor. Is this harmful to me only if I combine it with dextromethorphan and/or pseudoephedrine? Or is it harmful in and of itself if I have been diagnosed with CMT? I seem to be experiencing more flexibility in my left foot, and my hands seem stronger. Is this the accentuated reflexes mentioned in the warning? What is harmful about that if I am not using any cold medication?

I am currently taking Lisinopril for high blood pressure. It contains an Ace inhibitor and a diuretic. Is it okay for me to take? I have just started the second month.

To control hot flashes, I am using the Vivelle hormone patch.

My gynecologist had never heard of CMT. She is the one who prescribed the Effexor and Vivelle.

Thank you for this opportunity to ask questions from experts who know and understand this condition.

The Doctor Replies:

Venlafaxine (Effexor) is not known to worsen CMT or neuropathy in general. In fact, it is an emerging, but not currently approved treatment for neuropathy discomfort. None of the other drugs mentioned are known to worsen neuropathy or CMT, but Effexor could potentially interfere with the metabolism of dextromethorphan, but probably not in a clinically meaningful level. The interaction should be discussed with your physician, if the cold medicines are frequently used.

Dear Doctor:

I've been on Coumadin for 15 years and I've noticed changes in my PT/PTT recently. (I was a bit lax in having my blood drawn prior to just a couple of months ago). I have also been taking Neurontin and Pamelor for the past 7 to 8 months, since being diagnosed with CMT. I'm a 40-year-old woman. I was diagnosed with CMT after the birth of my daughter in July, 2004. Have you heard of any possible side effects from use of these drugs? Could I be onto something here?

The Doctor Replies:

Many people take the blood thinner warfarin (Coumadin) for a variety of reasons. The drug interferes with blood concentrations of many other drugs and this fact needs to be taken into consideration when dosing, but there are no suspicions that it causes neuropathy or worsens CMT. Both Neurontin and Pamelor are used to reduce the symptoms of neuropathy and are not known to worsen the underlying neuropathy.

Dear Doctor:

I am 54 years old with shortness of breath not related to heart, emphysema, bronchitis, or any other obvious reasons. I had pulmonary breathing studies done along with an extensive cardiac work-up.

Specifically, my question relates to the drug Macrodantin. For a period of two years, I was taking a daily regimen of this drug along with an increased dose when a urinary tract infection would flare up. So all in all, I've taken quite a bit. Last year, (while still on the drug), I developed severe shortness of breath which prohibited me from walking. It was actually my rheumatologist who figured out the Macrodantin might be the culprit. So, the drug was stopped and the breathing did improve. Yet, after being off the drug for a year, my shortness of breath continues, although certainly not as severe as it was while taking the drug. I have an upcoming appointment with a pulmonologist, and was hoping to be armed with a little more information prior to the appointment. Can you tell me how this drug adversely affects CMT patients or why?

The Doctor Replies:

Nitrofurantoin (Macrodantin) is an antibiotic used for a variety of problems, but more extensively in past decades for urinary tract infections. It has been associated with causing and worsening neuropathy, especially if taken on an ongoing basis; however, the nerves that control respiration are not usually involved, especially if the nerves to the limbs are not similarly affected. There is surprisingly little information about CMT patients using this drug on a chronic basis. The drug, however, is associated with other direct lung (pulmonary) toxicities of differing types and is likely the cause of your problem. A pulmonolo-

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

gist would be better informed about what to expect about recovery depending on the type of problem causing the shortness of breath.

Dear Doctor:
I have never been formally diagnosed with CMT, but I have the symptoms as I read about it and a recent examination by a neurologist seemed to confirm that I have it. Others in my family—brother, daughter, cousins, and aunts—have diagnosed themselves with it.

About 6 years ago I had prostate surgery and was previously on Androderm patches. In prep for surgery I had to stop the Androderm and since then I have noticed a marked weakness in my legs. Walking down a hill is a scary situation. I have asked my doctor to prescribe Androderm again for a while to see if it will help in any way.

Is the use of Androderm advised or not advised? Why?

The Doctor Replies:
Male and female steroids (hormones) of various types appear to affect a variety of normal nerve and muscle functions. Treatment with onapristone, a progesterone antagonist, has improved the neuropathy of the CMT1A rat, but has not been tested in humans yet. Testosterone (Androderm) has beneficial effects on muscle building, but also has numerous deleterious effects, including those on prostate tissue. The changes described are not surprising following the loss of the hormone treatment, but testosterone has not been adequately examined for safety and effect in CMT patients to recommend use for neuropathy. However, it is not something to avoid in a CMT patient, if indicated for another condition. As with any treatment with possible toxicity, the relative risks and benefits need to be weighed. The potential benefit may simply be adding more muscle and not improved nerve function. ✲

LETTERS

Dear CMTA,
Those that complain about cold hands and feet should have their thyroid checked. I was diagnosed with an underactive thyroid and had very cold hands and feet all the time. After going on Synthroid medication, I am feeling much better overall.

I also had very severe cramping in my legs and all over my body until I figured out that it was caused by dehydration. I never drank water. I was addicted to chocolate and did not realize that it was a diuretic along with wine and soda pop, all causing me very severe cramps. I can eat those foods just as long as I drink a lot of water afterwards.

—P.N.

Dear CMTA,
I am writing from the little island of Malta. I thought it would be a good idea to share with you two shoe brand names which I have found very helpful. They are Ecco and Pikolino shoes. Both brands manufacture flat shoes, very lightweight, with or without laces. I have CMT, as does my mother. We used to find it so frustrating to find a pair of suitable shoes. Now that these two brands are sold locally, our life has become easier.

—L.V.

Editor's Note: The Walk Shop offers shoes from both of these shoe manufacturers; visit www.walkshop.com or call (510) 849-3628 for a catalogue.

Dear CMTA,
I teach children 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 jar cookie mix 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 choice. Your charity was one of those chosen by the fourth graders.

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

—D.M.

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.

CMT PATIENT MEDICATION ALERT:

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
Leflunomide
Metronidazole/Misonidazole (extended use)
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
Infliximab
Isoniazid (INH)
Mefloquine
Penicillamine
Phenytoin (Dilantin)
Podophyllin resin
Sertraline (Zoloft)
Statins
Tacrolimus (FK506, Prograf)
Zimeldine (not in U.S.)
a-Interferon

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

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, 1C, 1D (EGR2), 1X, HNPP, 2A, 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

CMTA
2700 Chestnut Parkway
Chester, PA 19013
1-800-606-CMTA FAX (610) 499-9267
www.charcot-marie-tooth.org

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