



**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.

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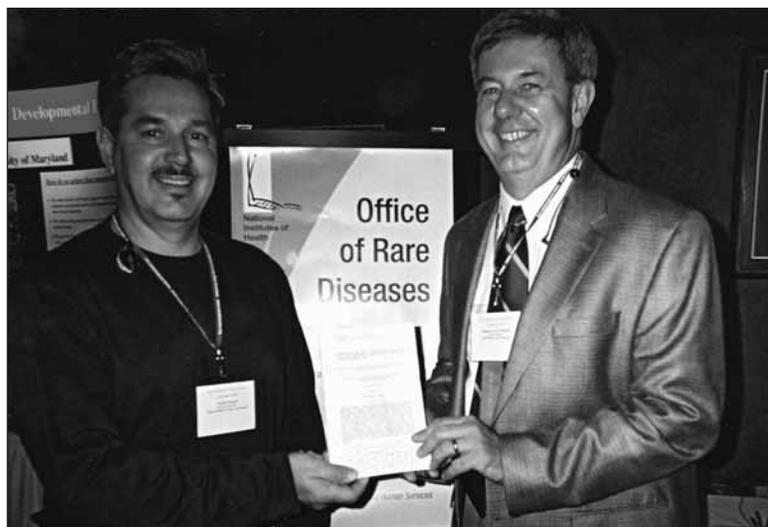
EMAIL CMTA AT:
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Together—One Voice The National Organization for Rare Disorders (NORD) Conference

BY PAT DREIBELBIS

On Thursday, October 14, 2004, Executive Director Charles Hagins and I flew out to Minnesota to attend the National Conference of Rare Disorders (NORD) conference in Bloomington. Both NORD and the CMTA were founded in 1983 and we have been a member organization of theirs for many years. The purpose of the annual conference is to bring together both patients and advocates of the hundreds of rare diseases that plague our country. NORD helps groups and individuals celebrate the promise of extraordinary medical and social advancements for people with rare disorders, and identify the areas of future concern that could inhibit or delay future advancements.

The opening address, "Understanding Research: Getting the Inside Scoop" helped the audience understand the difference between basic and clinical research. In some ways, the presentation was helpful in making everyone understand that gene research (how does the disease begin, what is the cause?) is always the first step. Clinical research, in which drugs or procedures are tested, always follows basic research and it can often



CMTA Executive Director Charles Hagins accepts congratulations from Dr. Stephen Groft for having CMT research language passed by Congress.

cost up to \$900 million to get from basic research all the way to a product being placed on the market. Human research, looking at efficacy, dosing, regimen, and how a drug or therapy might be administered, is considered phase two of research. Patients were reminded that drug companies are always financially driven and they must answer to their shareholders, so it is difficult to get "orphan drugs" and therapies to market.

From a researcher's perspective, Dr. Leslie Cooper of the Mayo Clinic explained that it is often difficult to find patients in sufficient numbers and that dedicated researchers need to collaborate when dealing with rare

disorders. Of course, he also stressed the difficulty in finding money for rare disease research and the fact that it often takes ten or more years to make clinical progress, which is frustrating for researchers and patients alike.

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A Different Approach to Bracing: The Silicone Ankle-Foot Orthosis (SAFO)

BY DANA SCHWERTFEGER

On September 24, Pat Dreibelbis and I traveled to Temple University in downtown Philadelphia. Dr. Howard Hillstrom, Director of the Gait Study Center at Temple, had invited Pat to attend a presentation by Dorset Orthopaedic, the British company that makes the silicone ankle-foot orthosis (SAFO), and I tagged along out of curiosity. As someone who has worn molded ankle-foot orthoses (MAFOs) for 25 years, I was interested in seeing whether SAFOs would enable me to walk as well as I do and be more comfortable than I am with MAFOs.

We entered the Gait Study Center, a long, narrow room with a raised platform along one wall and all sorts of cameras, computers, and gadgets, and were greeted by Dr. Hillstrom, whose area of expertise is the biomechanics of walking. Dr. Larry Newman, a podiatrist and Director of Temple's Foot and

Ankle Institute, joined us as well and provided helpful information about the structure of the foot and ankle.

While we waited for the people from Dorset, Bob Watts and Matthew Hughes, to arrive, we swapped stories about CMT and bracing with two patients from the Philadelphia area and another who had flown in from Seattle to be measured for SAFOs. Like me, Brian was in his 40s, had severe atrophy of his lower legs, and had worn MAFOs for years. The patients from Philadelphia were older and less severely affected. One wore MAFOs, but the other managed with an ankle brace and a cane.

We might have hoped for a better turnout, but we provided a fairly representative sample of CMT patients and having a small group had its advantages. During Matthew's PowerPoint presentation, we had the opportunity to ask numerous ques-



SAFO Brace

tions and then we all had a chance to try walking while wearing SAFOs.

Many of you know what MAFOs are, but for those who don't, I'll describe them first so you can see how different SAFOs are. MAFOs are made from semi-rigid plastic, usually polypropylene, that is vacuum formed to a positive cast of the lower leg. They extend from the upper calf, down the back of the heel, and under the foot, holding the foot in a slightly dorsi-

NORD

(Continued from page 1)

Other topics included "Surviving Your Doctor's Care," "Creating a Family Medical History," "Managing a Web Site and Database," and "Getting the Word Out."

NORD partnered with the National Institutes of Health,

Office of Rare Diseases to bring this conference to their member agencies. We were fortunate to meet with Stephen Groft, PharmD, Director of the NIH Office of Rare Diseases, to discuss the language on CMT research funding that was recently passed by Congress.

This conference was both encouraging and disheartening.

The CMTA seems to be moving forward in a very constructive manner, increasing its membership and making its presence felt in the research and government arenas. Other organizations, founded by parents with very sick children, struggle on with only twenty or thirty families dealing with the same frightening, often fatal, disease. ✱

CMTA MEMBERSHIP/ORDER FORM

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

	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 <input type="checkbox"/> English <input type="checkbox"/> Spanish		active members \$3 inactive members \$5	
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CMT Facts III		active members \$5 inactive members \$7	
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CMT Facts V		active members \$12 inactive members \$15	
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Check payable to the CMTA (US residents only).
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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.

flexed position. (Dorsiflexion is what you do when you pull your foot up toward your knee and plantarflexion is just the opposite, pushing the foot down and away from your knee.) MAFOs are secured by one or more Velcro straps, and require the patient to wear larger shoes to accommodate them.

Many patients say MAFOs are uncomfortable and create pressure sores where they rub against the ankle. I've tinkered a bit and made mine comfortable, so I don't have sores and can walk fairly well, but they're still not fun to wear on a hot summer day and they don't make much of a fashion statement when I wear shorts.

As we learned from Dr. Hillstrom, however, wearing MAFOs also affects the biomechanics of walking. Without MAFOs, both Brian and I walked with a high "steppage gait"—using our hip flexors to lift our knees high so that we didn't trip, and then flopping our feet down rather than setting them down in normal heel-to-toe motion. Wearing MAFOs allowed us to walk heel to toe, but Dr. Hillstrom showed us how the rigidity of the MAFOs forced our knees forward as we did. He also had us stand on a pressure-sensitive plate so we could see how wearing MAFOs also affected our balance and changed the way we distributed our weight while standing. We both tended to slump forward a little at the knee and lean into the Velcro straps, not exactly the most desirable posture because of the stress it placed on our backs. (I try not to stand still

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SAFO BRACE

(Continued from page 2)

for long periods of time, but now I know why my back hurts when I do.)

Bob Watts and Matthew Hughes then explained how SAFOs had been designed seven years ago for a CMT patient to make walking with braces more natural and comfortable. Like MAFOs, SAFOs are custom fitted, so negative and positive casts of the foot and ankle must still be made. Matthew demonstrated the negative casting procedure on Brian, and he made special note of bony prominences and other areas that would require special accommodation when the positive casts were made and the silicone sheets formed around them.

Unlike the semi-rigid polypropylene used in making MAFOs, the silicone is flexible and has a rubbery feel, almost like a hot water bottle. For cosmetic purposes, the silicone can be matched to a person's skin tone or made in a pattern for children. Since SAFOs only extend four to five inches above the ankle and have wrap-around Velcro closure instead of D-rings and straps, they are hardly noticeable. They can be worn with or without socks, while showering or swimming, with normal size shoes, and with heels up to an inch and a half in height.

Bob said SAFOs have a life expectancy of about two years,

but I suspect you'd reduce that if you did too much walking without shoes. The silicone is built up on the front of the leg and over the instep of the foot, but it's much thinner on the sides and bottom of the foot.

Cosmetically, SAFOs seem to be a vast improvement over MAFOs, but what about comfort and function? The pair I tried on weren't custom fitted to me, and I only wore them for about 15 minutes, so I can't real-



Brian, a patient from Washington State, was the guinea pig as he had his feet and ankles cast for the SAFO brace.

ly say much with regard to comfort. Brian has been wearing his for about a week now and says he's still getting used to the feel of the silicone; other people have said SAFOs make their feet sweat. Except for the toes and an area around the heel, SAFOs completely enclose the foot and ankle, so reports of sweating don't seem surprising. I don't

know what Dorset is doing to address the sweating issue, but they may be able to refine the manufacturing process to allow for additional ventilation.

I haven't seen Brian walk with his custom-fitted SAFOs, but I did notice an immediate improvement in his gait with the pair he tried on at the Gait Study Center. When my turn came, I wasn't sure the SAFOs I tried were going to provide enough support. After years of wearing MAFOs, the SAFOs seemed almost too flexible. I was able to walk fairly well, however, and my only problem was controlling my heel-toe motion so that my feet didn't slap. With MAFOs, I can easily walk several miles at a good pace before tiring, but I'd have to walk some distance in SAFOs before I could say whether I'd experience more or less fatigue. I also wasn't as steady on my feet while standing still, but Bob and Matthew said it would take about three months before someone was completely comfortable with SAFOs.

Dr. Hillstrom said the SAFOs appeared to result in improvements in our gait and posture, and he's applying to the CMTA for funds to do a formal study. Meanwhile, more information can be obtained from Dorset Orthopaedic. Their email is enquiry@dorset-ortho.co.uk and you can read about SAFOs and view video clips at two websites:

National Parks Service Offers Golden Access Passport

FOR CITIZENS OR PERMANENT RESIDENTS OF THE UNITED STATES WHO ARE BLIND OR PERMANENTLY DISABLED

The Golden Access Passport is a lifetime entrance pass to national parks, monuments, historic sites, recreation areas, and national wildlife refuges that charge an entrance fee. The Golden Access Passport admits the pass signee and any accompanying passengers in a private* vehicle if a park has a per vehicle entrance fee. Where a per person entrance fee is charged, the Golden Access Passport admits the pass signee, spouse, and children.

The Golden Access Passport also provides a 50% discount on Federal use fees charged for facilities and services such as camping, swimming, parking, boat launching, and tours. In some cases where use fees are charged, only the pass signee will be given the 50% price reduction. The Passport is nontransferable and does NOT cover or reduce special recreation permit fees or fees charged by concession people. A Golden Access Passport must be obtained in person at a



Federal area where an entrance fee is charged. It is available only to citizens or permanent residents of the United States who are medically determined to be blind or permanently disabled. You may obtain a Golden Access Passport by showing proof of medically determined permanent disability, or eligibility for receiving benefits under Federal law.

The Golden Access Passport can only be obtained in person at a Federal area (national park, historic site, wildlife refuge, etc.) where an entrance fee is charged.

If your Golden Access Passport is lost or stolen, there is no mechanism for replacement. A new pass must be obtained.

You can use our “Visit Your Parks” portal on our website (www.doi.gov) to locate a National Park Service area near you. We suggest that you telephone your local National Park Service site to inquire about pass availability and what documentation you need to provide before traveling. *

* A private vehicle is defined as any noncommercial vehicle that is being used for private recreational purposes.

www.dorsetorthopaedic.co.uk and www.safp.eu.com.

Although Dorset has been making SAFOs in the U.K. for seven years, SAFOs are not currently manufactured in the United States. A qualified orthotist can, however, measure and cast you and send the negative casts to Dorset. Dorset

will ship the SAFOs to your orthotist approximately 10 days after receipt of the casts. As to price, we are informed by Dorset that SAFOs have been fitted by orthotists in the U.S. at a cost of \$4,500 a pair, but are available in the U.K. from Dorset at £1,232 (\$2,254) a pair. The reason, Bob said, is

that medical costs are generally lower in Europe than in the U.S., but what we don't know yet is how insurance providers will respond to requests for SAFOs at \$4,500 a pair, or even at \$2,254, so we'd be interested in hearing from anyone who tries to obtain payment for SAFOs. *

Books Worth a Look

BY PAT DREIBELBIS

HOW TO SURVIVE YOUR DOCTOR'S CARE

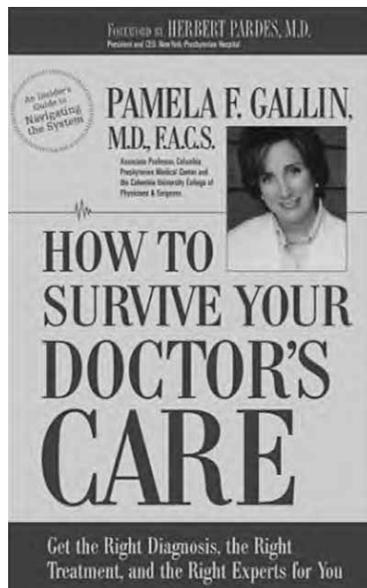
By Pamela F. Gallin, M.D.,
F.A.C.S.

At the NORD (National Organization of Rare Disorders) meeting in Minneapolis, MN, Dr. Gallin spoke on the topic, "How to Get the Doctor You Need and the Care You Deserve." As a physician at Columbia Presbyterian Medical Center, Dr. Gallin has chosen to look at the medical system from the inside out.

She discusses the problems in finding doctors who are aware of the particular rare disease that the patient represents and writes that diagnostic mistakes often occur because the doctor has never seen the problem before. To that end, she suggests finding the "guru" for your disease and having him build a team with your local specialist. Most importantly, she urges patients to ask questions and not be shy.

In her book, she amplifies these points and provides examples of what can go wrong and how to navigate the system. She discusses choosing the right hospital, becoming your own best advocate, deciding on surgical procedures and communicating effectively with doctors and nurses.

The book is published by LifeLine Press (ISBN number: 0865261200). It should be available at most larger book stores.

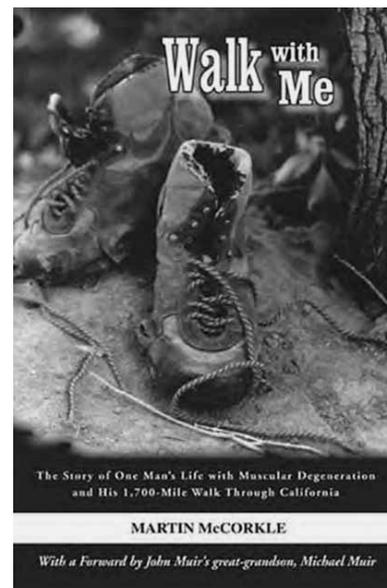


WALK WITH ME, THE STORY OF ONE MAN'S LIFE WITH MUSCULAR DEGENERATION AND HIS 1,700-MILE WALK THROUGH CALIFORNIA

By Martin McCorkle

Books written by people with CMT are rare enough that I would be tempted to read them no matter what the topic might be. When I was approached by the author to read a book about someone who hikes, I thought the book would be interesting, but not likely to appeal to me in any real sense. I was *very* wrong. This book is such an interesting and compelling story, not just of what the experience of hiking and viewing nature might hold, but more so, a story of a man's life and how he has handled various challenges, disappointments, and successes.

The book begins with a



challenging hike scenario, but quickly flashes back to his experience with his first EMG (electromyogram) and the pain of needle pricks and electrical stabs. From that point on, the book alternates between memories of dealing with CMT as a child, young adult, and current 50-something and the love of nature which has permeated Martin's life from childhood on.

He says in an early chapter, "Kids can be cruel. And they were from time to time. But my memories of dealing with CMT as a child are mostly filled with astonishing acts of kindness and understanding from my friends and my school."

As the book progresses, so does Martin's family life, his ministry and the problems with CMT. At times, hiking was forsaken altogether, but the pull of the Pacific trails always stayed with him. In dark times, he

The Annual Appeal... You are the CMTA!

2004 is quickly drawing to a close. The end of the year is always a time for reflection. This year, we are pleased to be able to report on many successes as a result of the continued support of our membership through the annual appeal.

For the first time, we successfully lobbied to have Congress recognize the importance of CMT research and include report language calling on NIH (National Institutes of Health) to consider and report on ways to increase funding for CMT research.

We redesigned our newsletter, *The CMTA Report*, to make it easier to read and have worked to include more quality-of-life information. We've also redesigned our brochure, *What is CMT?*, and expanded our Medical Alert list to make it more useful. Those publications are available now for our members.

We now have a part-time employee who is following up with people who request packets of information to see if he can help them further and to ask them to become paying members so our services can continue and expand.

We continue to support the CMT North American Database and to help enroll patients so that it will serve CMT researchers now and in the future.

The end of the year is also a time to compose a "wish list" for the oncoming year, and we are excited about our plans for the future.

We want to fund the first clinical trials in North America on ascorbic acid and we hope to call for participants in that study through our newsletter and website.

In May of 2005, we will be sponsoring the Second Annual Symposium of the North American CMT Consortium where researchers

meet to discuss their successes in CMT research and avenues for further study.

We plan to launch an entirely new website with more information and many new features which will benefit our membership. Navigation will be improved and archived newsletter articles will allow members access to far more information.

In conjunction with the newly designed website, we are launching a children's initiative which plans to design materials for use in schools and which will establish a parents' network and resource center.

Both our successes in the past and our dreams for the future depend on *you*. Your support of our annual appeal will allow us to improve the lives of people with CMT. Please continue to support us generously.

—Charles Hagins,
Executive Director

experienced too many falls and bloody knees. Still, he made bargains with himself about his hiking and his CMT.

One of my favorite sections of the book is the one in which he almost, but not quite, concedes to the progression of CMT and gives up his dream of completing the Pacific Crest Trail. He admits that it would be easy to give in and just enjoy his home, his family, and his sofa, but then he realizes that only one thing

could give him the desire to defy CMT again—this need to go back to Lookout Rock and walk the rest of the way to Oregon on the Pacific Crest Trail. He writes, "The Pacific Crest Trail is more than scenery—she is one of my life's few true passions... If I quit living, what good am I? For every year that I defy CMT, I have another active year with my loved ones. God placed a love for this trail in my heart for such a time as this. I needed a strong love to

work against a strong foe."

I won't divulge how this book ends, but I will say that reading it is well worth your time and money. I was uplifted and deeply satisfied, when, with Martin, I came to the end of his journey.

The book can be purchased at publishamerica.com for \$16.95. The ISBN number is 1413730434. You can also reach Martin McCorkle at his website, www.martinmccorkle.com *



GIFTS WERE MADE TO THE CMTA

IN MEMORY OF

Betty L. Chow

Aileen Arakawa
 Burbank INSS Staff
 Vern & Imgard Burling
 Gladys Chow
 Glenda Chow
 Jane Chow
 Lorraine Chow
 Mavis Chow
 Stanley Chow
 Allen & Martha Chun
 Dorothy Chun
 June & Laurel Colton
 Tim DeFiesta
 Anna Fong
 Tak & Judy Furumoto
 Hajime & Yaeko Hironaka
 Gary & Amy Ho
 Butch & Patti Hong
 Dick Hong
 James & Susan Hong
 Janet Lum Hoy
 Jean Kaung
 San & Terry Kitamura
 Gene & Jade Lew
 Lifescan – Johnson & Johnson
 Josie Mercado
 Debbie Nakasato

The CMTA was saddened to learn of the untimely passing of Betty Chow who ran fundraisers for the association and was a tireless worker on behalf of CMT awareness. Our deepest sympathies go to her family and friends.

Betty L. Chow (continued)

Craig Ng
 Fred & Sharleen Ozawa
 Pearl & Larry Snodgrass
 Eleanor Telemaque
 Randy & Valerie Wong
 Darrell & Ava Yap
 Darren & Frances Yap
 Dave & Millie Yap
 Laura Yuen
 Rose Yuen
Virginia Coombs
 Deborah Militello
Frank Eckman
 Frank & Debbie Tamburello
Kenneth Fleeman
 Sanders Tools & Supplies, Inc.

Robert Herron

Scott, Jim, Bonnie & Carolyn

Peggy Jones

Kettle Creek Church of Christ

Wallace Sheldon

Howard Martin
 Mr. & Mrs. Brian Ponting

Sayre Thompson

Ernest & Mary Florestano

Thelma Yampolsky

Harriet & Frank Weiss

IN HONOR OF

Yohan Bouchard

North Avenue Express Deli

Dr. Robert Chetlin

Marilyn Freed

Gail Feeney-Coyle

Marian Bakken

Herbert Holly

Dorothee Knowles

Lauri & Jonathan Ley

Jordan Ley & Brianna D'gata

Joe Seidler

Harriet & Frank Weiss

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: _____



LIVING WITH CMT

Getting the Ball Rolling

BY DANA SCHWERTFEGER

Since I began working part time at the CMTA several months ago, I've begun calling people a few weeks after sending them an information packet. It seemed like a good idea to follow up and see if people had additional questions or needed assistance, and I've been living with CMT for 30 years so I was fairly confident I'd be able to answer everyone's questions.

I guess I don't have to tell you how long it took me to realize that there was more about CMT that I didn't know than there was that I did know, so it's been quite a learning experience!

But it's also been very rewarding. Many of the people I've spoken with are just learning about CMT, and they're as frightened and confused as I was when I was first diagnosed. They're worried how fast their CMT will progress and how severely they will be affected. No one can tell them that, of course, but many are relieved just to be talking with someone who actually has the disease and who is functioning pretty well after 30 years.

As amazing as it sounds, I've also spoken with a number of people who've had CMT for years but have never spoken with anyone else who has it. No one ever told them about the

CMTA, and they've had to cope on their own. They've been very resourceful and I often learn as much from them as they do from me, which is a point I'll return to in a moment.

Overall, though, speaking with parents of children with CMT has proven to be the greatest challenge. I'm not a parent myself, so I don't have any direct experience to draw on.

Each of you has a wealth of information that can benefit us all.

I'm always looking for ideas, however, so at some point in the conversation I often ask if they have any suggestions about how the CMTA could help.

Rebecca, the mother of a seven-year-old, said that a booklet written specifically to educate teachers, school nurses, and gym instructors would be a good idea. I asked if she'd be willing to help put one together, and that got the ball rolling. Over the next few weeks, I kept that in mind while talking with other parents, and before I knew it I had a group of eight parents willing to help.

And not just with that booklet. Stacia, whose daughter is six, suggested a booklet

explaining CMT to other schoolchildren. That's a great idea, too. Then Cheryl told me about all the resources she tapped in her state to get help for her twelve-year-old son. Wow! How great would it be to have a list of resources like that for each state? We could put those on the website where all members of the CMTA could access them.

You can see how quickly things were getting out of hand, but the light bulbs just kept coming on. What about a network—a place for parents (and kids) to speak with each other and exchange ideas? That could be worked into the redesign of the website, too, but why stop there? Why not host an annual gathering where the parents and kids could meet each other, have a good time, and maybe attend workshops or seminars?

Churning out ideas is easy. Translating them into reality takes time and plenty of hard work. That's where you come in. Remember I said I learned as much from some people as they learned from me? Well, *you* are the CMTA and now, more than ever, we need your input.

Each of you has a wealth of information that can benefit us all. Let's talk just about state

(continued on page 15)



SUPPORT GROUP NEWS

Do you live in a location that is not currently served by a CMTA support group? If that is a situation you would like to remedy, you might have the solution right in your own hands. The support group liaison, Bob Budde, can offer invaluable insight into starting and running a support group since he started and led the Kentucky group for many years. You can reach Bob at 1-859-255-7471. Or, you can call the main office at 1-800-606-2682 and receive the “How to Start a Support Group” information. There is no special skill required in order to run a successful group other than commitment. Our group leaders are younger and older, with CMT themselves, or having a family member with the disorder, experienced in group dynamics or complete novices and all enjoy the same successes. So, rather than waiting for someone else to start a group that could serve your needs, be the leader and get something organized that will help you and others in your area.

Pennsylvania—Johnstown Area

The September Johnstown CMTA support group meeting was held at The John P. Murtha Neuroscience and Pain Institute in conjunction with “September is CMT Awareness Month.” Attendance was fantastic with over 30 attendees (lots of new members) We now have over 80 members in our group.

Two TV stations covered the event—the NBC affiliate,



Members of the Johnstown Support Group examine various AFOs (braces) to compare function and comfort.

WJAC-TV and the Fox affiliate, WWCP-TV. The coverage included a filming of the meeting and interviews with Jeana Sweeney, the support group co-leader. The film aired on the 6:00 and 11:00 PM news.

The primary speaker was Dr. Nassr, Chief of Neurology, The John P. Murtha Neuroscience and Pain Institute, who gave an excellent presentation and did a great job of answering questions.

The meeting also included a preview of the “Conemaugh Connection with Amy Bradley,” a half-hour TV special by The Conemaugh Health System, featuring CMT. The show aired in prime time, 7:30 PM, and four other times on the NBC affiliate, WJAC-TV. The program also airs on the public access channel periodically. This is an excellent and moving piece and has really helped raise CMT awareness in our area. We want

to thank The John P. Murtha Neuroscience and Pain Institute (in particular Barb Duryea and Jan Goodard), the Conemaugh Health System, and Amy Bradley for this program, the work they do, and their excellent CMT clinic.

This disease is not rare and it is amazing how raising awareness brings people with CMT or those who think they have CMT forward. The group puts a lot of effort into raising awareness here and it seems at every CMT event or presentation someone has or knows someone else who has CMT. Anecdotally, Jeana’s husband is a mail carrier and has identified two families with CMT on his route.

JD Griffith and Jeana Sweeney also give CMT awareness PowerPoint presentations to civic, church, and service groups (or any group with a screen or wall) and we travel in the area to continue our outreach. ❄

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

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—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: Jeanie Porter, 503-591-9412
Darlene Weston, 503-245-8444

Email: jeanie4211@hotmail.com or
blzrbabe@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,
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 have Crohn's disease and have taken Remicade (infliximab) for five years. It seems to me that since I started taking this treatment, my CMT symptoms have worsened. There are several warnings on the drug and it is not recommended for people with nervous system disorders. Can you look into this for me?

The Doctor replies:

Last month, I would have said this drug is of little concern. The preliminary studies being done for drug approval showed some patients with neuropathy, but it was not clear if the drug had any relation to the problem even though it is listed as an association in the PDR (Physician's Desk Reference). There are other neurological concerns in the central nervous system. Other than the PDR, there was

no medical literature I could find to support the neuropathy risk. However, an article was just published (11/9/04) from Germany that found two patients with new neuropathy that probably, but not definitely, was caused by the drug. We will add the drug to our list in the minor to uncertain risk. For the patient, I would advise that it is a possible, but rare, effect that should be addressed with his/her doctor. Other reasons for CMT worsening, such as the Crohn's disease, should also be considered before changing therapy.

Dear Doctor:

I've read many recommendations to take antidepressants to alleviate some CMT symptoms. But other people say that people with CMT should not be on medications like that. Do you have an opinion?

The Doctor replies:

Pain in CMT is frequent and there are different causes that produce pain, including calluses in the pressure areas of the feet and arthritic pain. The last type responds to anti-inflammatory medication. Antidepressants are good for dealing with the burning neuropathic pain. They are safe in CMT patients. You should consult with your neurologist to find the best treatment for your specific pain.

Dear Doctor:

Are people with CMT prone to reactions from anesthesia when having surgery?

The Doctor replies:

There is no good literature that I am aware of that suggests anesthetic agents are a risk for CMT patients. However, prior to receiving anesthesia, it is proba-

Some Facts About Pain

Pain syndromes are not uncommon in CMT. Age at onset, length of time with CMT, or degree of disability plays no part in distinguishing people with pain from those who are pain-free.

If you feel pain, it's important to communicate that information to your physician. Describe the symptoms and the degree of severity you feel as clearly as possible because treatments vary. Treatment can help prevent pain from taking over a person's life.

Pain that occurs as a result of nervous system disease is different from the "good" pain that serves to protect the body from further injury. There are nerve endings sensitive to pain all over the body, which sound an alarm to the brain when something is harming it or about to harm it. The brain normally responds so quickly that a person has already withdrawn the fingers from the hot stovetop before realizing consciously what happened. This is functional pain at work, preventing serious injury.

In more serious events, such as a sprained ankle or a broken bone, pain sounds a general alarm designed to make the individual rest and let his body heal. This, too, is functional pain with a purpose.

In contrast, "chronic" or recurrent pain—especially pain that occurs because of disease or injury to the nervous system—serves no useful purpose. It robs sleep, saps energy, dampens mood, and curbs appetite.

It is important for you and your doctor to distinguish

bly a good idea to undergo pulmonary function testing just to make certain that you will not have problems coming off the respirator after surgery.

Dear Doctor:

I am 49 years old, female, and have CMT. I am waiting for a blood test to see which kind of CMT I have. In my readings about CMT, I see that some people complain about fatigue. Some doctors say that CMT is not the cause, but so many people with CMT complain about it that I wonder if there is some direct connection. Is it possible that CMT makes a person low on iron and I should take iron supplements to alleviate anemia.

The Doctor replies:

Fatigue may be produced if the symptoms of CMT are severe enough to produce a laborious gait. Additionally, the difficulty of performing day-to-day activities is part of the story. CMT is a

genetic defect and has nothing to do with iron or anemia. A blood test can determine if you have an iron deficiency, in which case, an iron supplement may be helpful.

Dear Doctor:

I recently was prescribed hydroxychloroquine for an autoimmune disease. I noticed in The CMT Report that chloroquine is listed as an "Uncertain or minor risk". Would this level of risk also apply to hydroxychloroquine? I am 53 years old and was diagnosed with CMT many years ago.

The Doctor replies:

Hydroxychloroquine (Plaquenil) is associated in rare cases with toxicity to muscle and even less commonly with peripheral neuropathy. Only a handful of cases among the tens of thousands or more treated have been described to have peripheral neuropathy. It is not clear if CMT patients are at undue risk. The question of toxicity should be raised if weak-

ness develops or worsens soon after starting the drug, especially in shoulder and hip girdle muscles, but the nerves and muscles of most treated patients appear to be unaffected.

Dear Doctor:

I am a 42-year-old man diagnosed with CMT with severe foot drop and foot contractures in my right foot only. I also have right patella problems because of foot drop. Would heel cord lengthening help foot drop and patella problems?

The Doctor replies:

It is always difficult to answer a question like this without seeing the patient and evaluating him. In general, foot drop in CMT is a result of muscle weakness secondary to damage of the peroneal nerve and is independent of the heel cord, Achilles tendon, or the patella. Therefore, in most cases, a better approach is good ankle-foot orthotics rather than surgery on the tendons. *

between CMT pain and other kinds of pain. Having CMT doesn't make you immune to headaches, cramps, arthritis, or low-back pain. You and your doctor need to consider the nature and source of your pain. The main requirement for effective pain control is proper diagnosis. People must be "in tune" with their bodies, so they can explain or describe symptoms in a way that will aid diagnosis.

Burning, aching, prickling, or "pins and needles" may be chronic pain rather than acute pain. These pains are sometimes treated with gabapentin (Neurontin) or with antidepressants

such as amitriptyline (Elavil) because such agents modify how the nervous system reacts to pain. Sometimes, treatment can include wearing a pressure stocking or glove to convert the sensation of pain to one of pressure. Warm compresses to the skin can convert the sensation of pain to one of warmth.

Chronic back pain can be caused by pressure on the body caused by immobility, incorrect use of mobility aids, or stress on the body from the struggle to compensate for walking problems. An evaluation to pinpoint the problem is essential. Treatments may include heat, mas-

sage, ultrasound, and physical therapy.

New facts about chronic pain and its consequences have an effect on how medicine is being practiced now. Pain is being regarded now as the "fifth vital sign." When a standard physical exam is being conducted, a physician will check the vital signs—temperature, blood pressure, heart and respiration rates, and now "pain level" as that fifth sign.

Acknowledging that pain should be documented and treated is a major step in overcoming the all-too-common belief that pain is all in your head. *

WRITE TO US!

Pat Dreibelbis, Editor
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The CMTA reserves the right to edit letters for space.



LETTERS

Dear CMTA:

Approximately a year ago I read an article in your newsletter that called my attention to Ortho Rehab Designs in Las Vegas. My current age is 62 and over the past twenty years I have progressively lost my calf leg muscles and developed severe drop foot. Accordingly, I did not walk very much because it was a huge task and took a lot of effort. I also could not stand upright to carry on a conversation with anyone since I had to steady myself by holding onto a fixed object and was using a walking cane. I had tried some AFOs, but never wore them since they just did not do the job of assisting me in walking.

After reading your article on Mitch Warner and Ortho Rehab Designs, I visited his website and then telephoned him to get additional information. This led me to travel from San Jose, CA to Las Vegas to visit Mitch for his evaluation of my situation. Mitch was very patient and convinced me that he could help me.

After the process of casting my legs and the building of the Orthos that fit my legs and feet perfectly, I tried them on for the first time. I could not believe the difference. I was able to stand immediately without holding onto someone or some fixed object. It has been a little over a year now since I first received the Helios Ortho's and my calf muscles have gotten stronger and I actually am getting some feel-

ing in my feet. I have been walking a lot more than I did before and that has led to stronger muscles in my legs. My younger wife has trouble keeping up with me when we are walking. I go to socials and

sporting events now without worrying about the crowds.

I owe so much to Ortho Rehab Designs. You can visit their website at www.orthosignslv.com

—E.M., San Jose, CA

CMT Database Participants Needed

The CMT North American Database is a collection of information on people with all types of CMT.

This information is critical to understanding more about these conditions and the ability of researchers to develop treatments for people living with Charcot-Marie-Tooth (CMT) disease. The information collected will also be helpful in determining prognoses and finding the best time in a person's life to apply treatments for their CMT. The main purpose of the database is simple; to help speed the pace of research so that real treatment options or a cure for CMT can come about *as soon as possible!* A cure will only happen if many people are willing to participate in research.

Participants with CMT are asked to fill out a few questionnaires about their medical and family histories. The database staff is available to help with these forms as needed. Researchers will have access to nonidentifying information in the database but all personal information is kept strictly confidential.

Research is helping us to learn more about CMT everyday. Our hope is that through the efforts of families with CMT we will someday find a cure. The CMT North American Database is essential to this goal. Be proactive in finding a cure for your disease: join the CMT North American Database! Database forms are available online at: http://www.med.wayne.edu/neurology/clin_programs/Labs/CMT/index.htm Also, please feel free to contact Carly Siskind at (313) 577-5273, or email her at csiskind@med.wayne.edu. *



Dear CMTA:

I have CMT Type 2. I am male, late 50's, height and weight proportionate, in generally good health, with moderate CMT symptoms. I recently got a kidney stone and an x-ray for that showed I had thin bones in the back, hip, pelvic area. So they did a bone density scan and said I have osteoporosis. The combination of osteoporosis and kidney stones suggests my calcium levels are screwed up, probably from a problem in a parathyroid gland which is the control center for calcium levels in the blood and bones. So I did some research on parathyroid in preparation for seeing an endocrinologist about that.

Calcium plays a big part of nerve conduction, muscle cramps, bone and joint aches, all of which are part of CMT. So, the hope is that by removing the offending parathyroid gland, the calcium will return to normal, the bones will get stronger and some of the other CMT symptoms will get better. And, if I ever fall from the CMT weakness, I have a better chance of not breaking any bones. Because right now if I

fall from CMT, there is a high chance I will break a leg, hip, wrist, or my back, all of which would be very difficult to recover from with the CMT.

How's that for toe bone connected to the foot bone, foot bone connected to the ankle bone, ankle bone connected to the leg bone.....

—B. E., e-mail

Dear CMTA,

Since reading your Letters column in the latest edition of *The CMTA Report*, I found one reader's recap of his diagnosis quite interesting. E.H. of Oakview, CA, complained of his doctors not being able to correctly diagnose his symptoms. I am a 67-year-old female who had a diagnosis of CMT disease after the birth of my third child in 1970. After visiting several doctors in my area of San Diego, CA, I finally found a neurologist in La Jolla, CA, who correctly diagnosed it by just looking at my thumbs. Subsequently, I found my way back to the MDA clinic at Children's Hospital in San Diego and received complete care for my CMT. It was a year-long search.

I am not surprised that E.H. did not find the diagnosis years ago. But, now there are many wonderful physicians here in California who are not only treating patients with CMT, but also doing research such as that at the Muscle Disease Clinic at UCLA.

One interesting note to E.H., I am contemplating knee replacement surgery after an unsuccessful arthroscopic procedure due to a torn meniscus. This minor surgery left me on crutches for three years. I am very skeptical of any surgery that may weaken an already compromised knee.

Thank you for the wonderful information you report in the newsletter.

—K.D. San Diego, CA

Dear CMTA,

I registered for IGive.com following your directions and feel so good about a portion of my Internet shopping going back to the CMTA, which has been so helpful to me and my family over the years. I hope others will do the same thing. The small percentages really do add up.

—C.H. Wallingford, PA

LIFE WITH CMT

(Continued from page 9)

resource lists, for example. I could spend a year trying to compile a comprehensive list for just one state, but if each of you were to take a moment to jot down the agencies and organizations you've dealt with and found helpful, think how quickly we could amass those lists.

Let me be even more specific. If you know of an agency, organization, or other resource for parents or children in your state, send a description or link to us at the CMTA. You can mail it to 2700 Chestnut Parkway, Chester, PA 19013, or you can email it to CMTAssoc@aol.com. While I'm at it, we're also looking for psychological help—to review and contribute

to the booklets, that is. So, if you know a child or school psychologist or other school administrator willing to get involved, please let us know.

Last, but not least, if you are a parent and interested in more information about participating in CMTAParentsNet, send us your contact information, preferably an email address. Thanks. ❄

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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 (ddl, Videx)

Dichloroacetate

Disulfiram (Antabuse)

Gold salts

Leflunomide

Metronidazole/Misonidazole

(extended use)

Nitrofurantoin (Macrochantin,

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-Fluorouracil

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



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