



**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.

# CMTA Hosts Second International Charcot-Marie-Tooth Consortium

BY PAT DREIBELBIS

From July 18, 2007 through July 20, 2007, one hundred and twenty-one of the world's finest researchers and clinicians gathered in Snowbird, Utah, to share their findings and to discuss the CMT research that is on-going throughout the world. It was a collaboration of the largest number of participants that the consortium has ever had. One of the reasons for the spectacular turnout was the fact that the consortium piggy-backed its meeting onto the Peripheral

The July consortium had the largest number of participants ever.

Nerve Society meeting which directly preceded it.

The meeting was begun by an introductory welcome from Dr. Michael Shy who had organized the scientists and their presentations. Without his work, the meeting could not

have happened, and without his choice of speakers and posters, it could not have been as well-organized or as informative as it was. Following his opening remarks, the first session was centered on the topic of new loci and genes that have been found to cause CMT. The majority of the discoveries involved autosomal recessive forms.

On the second day of the meeting, Klaus-Armin Nave, Director of the Department of Neurogenetics, the Max Planck Institute, Goettingen, Germany, *(continued on page 4)*

## CMTA BOARD

**Patrick Torchia**  
Chairman and President

**Robert Kleinman**  
Treasurer/President Elect

**Phyllis Sanders**  
Secretary

**Gary Gasper**

**Patrick Livney**

**Steve O'Donnell**

**Elizabeth Ouellette**

**Jason Steinbaum**

**Michael Shy, MD**

Chairman,

Medical Advisory Board

**Robert E. Lovelace, MD**

Chairman Emeritus,

Medical Advisory Board

**Jack Walfish**

Board Member Emeritus

**Charles F. Hagins**

Executive Director

**EMAIL CMTA AT:**

[info@charcot-marie-tooth.org](mailto:info@charcot-marie-tooth.org)

## The CMTA Awards Three-Year, \$300,000 Grant

The recipient of this year's three-year grant is Dr. Michael L. Garcia, Assistant Professor, at the University of Missouri, Columbia. His work is entitled "Mechanisms of disease pathogenesis in neurofilament-linked Charcot-Marie-Tooth disease."

The objective of this study is to characterize the role of NF-L (neurofilament light) mutations in the pathogenesis of CMT2E. The study group will generate two lines of mice expressing different mutations within different

functional regions of NF-L to provide a direct test of the link between NF-L mutations and CMT2E as well as provide insight into the variable onset and severity of the disease. Functional consequences of this form of CMT, such as abnormal gait, delay of heat response, slowed conduction velocity, and muscle wasting will be analyzed. These mice will provide the first animal models of CMT2E as well as understanding of the pathophysiology of neurofilament-linked

diseases such as amyotrophic lateral sclerosis (ALS).

Dr. Garcia will begin his project on January 1, 2008. \*

## INSIDE:

<b>Scooter Winner</b> .....	<b>2</b>
<b>Wayne State, Day 2</b> .....	<b>8</b>
<b>Quality of Life Issues</b> ...	<b>12</b>
<b>New Patient's Guide</b> ...	<b>17</b>

# Mobility Scooter Giveaway: And the Winner Is...

BY ELIZABETH OUELLETTE

**A**s I was descending the elevator of the Sears Tower in Chicago, where we were to meet Carol Simpson, the first recipient of our motorized scooter giveaway program, my heart was pounding with curiosity and excitement. After reading the recipient criteria for our mobility scooter offer, I realized that this was no small matter, as the winner would not only be offered a scooter, but more importantly, the prospect of independence and a sense of unwavering freedom from everyday limitations and barriers.

Not quite knowing what to expect, representatives of the CMTA, including Board mem-

bers, walked into the given room, where the air was electrified with anticipatory tension, animation, and wonder. Carol, the only member of her family to have CMTA, was sitting in a chair, smile on her face, braces on her legs, and crutches by her side. She was dutifully surrounded by many family members who came to witness this spectacular event and encourage Carol with their support.

I happily met Carol's mom Doris, her 2 sisters Patti and Gail as well as her niece, Erin, who lives in the Chicago area. As introductions were being made, I had to breathe deeply to keep my emotions at bay. I discovered that Gail had purpose-

fully flown in from Texas to be at Carol's side for the big day. Once Gail arrived, Carol's family settled into the car and began the 4-hour road trip to Chicago.

Up until this point, Carol's mobility had been extremely limited, mostly due to the effects of CMTA, complicated by other medical conditions. Although she does own a self-propelled wheelchair, she still needed a second party to push the chair when any type of distance was to be traveled. Despite her family's help, the time and energy it took to go to a mall, for example, was uncomfortably lengthy. For instance, they first had to wait for the wheelchair-accessible van to come and pick her up, then once at the mall, a family member would wheel her in, where she would use one of the mall's motorized scooters. After her shopping was completed, she returned the scooter, got back into the wheelchair, and waited for the van to pick her up and bring her home. It was an exhausting expedition for everyone involved!

So, once Carol was officially awarded the scooter, and after much congratulatory applause, hugs and cheers, she decided to sit on it and try it out for herself. While the Scootaround representative, a very kind and warm gentleman, showed her the controls and how best to maneuver her new form of transit, Carol's face was beaming radiantly.

**A representative from Scootaround and members of the CMTA Board of Directors awarded the first scooter to Carol Simpson.**



In no way was this event trivial; it was, in every sense of the expression, a dream come true.

As she tried out the forward and reverse maneuvers, I looked around the room and noticed that every single person was very deeply touched by Carol's gratitude, joy, and contagious opti-

and our partnership with Scootaround, Inc., this motorized scooter would concretely change Carol's inclusion and participation in the world, beginning that very moment and from then on.

I am extremely proud to be part of this wonderful organiza-

note, if you or someone you know is interested in applying for our next mobility scooter giveaway, please follow the instructions outlined within this newsletter or go to our website at: [www.charcot-marie-tooth.org](http://www.charcot-marie-tooth.org)

To close, I would like to extend a special note of thanks to our Executive Director, Charles Hagins, who embraced this program wholeheartedly right from the outset. I also would like to extend my appreciation to Dana Schwertfeger, for all his hard work in making this opportunity a reality, not only for Carol, but for all those who will follow in her footsteps. I have never, ever, met such a special group of people, so devoted and committed to a specific cause, as those who make up the CMTA. From my heart, I thank each and every one of you for relentlessly striving to improve the lives of those affected by Charcot-Marie-Tooth disorder. \*

This scooter will concretely change Carol's inclusion in the world.

mism and enthusiasm. Tears were welling up in everyone's eyes (of course, mine were already streaming down my face), overcome by the satisfaction and fulfillment of having the opportunity and the means of making one person's life with CMT a bit easier and a little less challenging.

Thanks to the CMTA, the Clarence Vincent Foundation,

tion for many, many, reasons. As you are reading about what CMTA is accomplishing with your generous donations (vitamin C trials, accelerated research plan, office expansion to Chicago and California, growth of support groups, to name a few), know that we are also devoted to improving the lives of those with CMT today, right now, in the present. On that

### Next Scooter Giveaway: November 2007

We are now accepting applications for the November, 2007, scooter giveaway. If you have already submitted an application and your information has not changed, we've kept your application on file and you do not need to resubmit. Otherwise, your application must be postmarked no later than October 15, 2007, and addressed to:

Scooter Giveaway  
CMTA  
2700 Chestnut Parkway  
Chester, PA 19013

To obtain an application, you may write to the above address, call 1-800-606-2682, email [scooter@charcot-marie-tooth.org](mailto:scooter@charcot-marie-tooth.org), or visit us on the web at <http://www.charcot-marie-tooth.org/scooter.php> \*



## CMT CONSORTIUM

*(continued from page 1)*

presented the Ann Lee Beyer Plenary Lecture on “Axon-glia interactions and demyelinating diseases.” The plenary address is named for former Board member and Chairman of the CMTA, Ann Lee Beyer, who fought tirelessly to bring researchers together to work for the common goal of finding a cure for CMT.

The rest of the morning was given over to researchers who were working on the cell biology of CMT. After lunch, researchers spoke on their discovery of unusual phenotypes and genotypes of CMT. Professor Garth Nicholson, Australia, posed the question of why the hereditary neuropathies are classified in such a confusing manner. He finds the continuing use of letters and numbers to be unnecessarily complex and would like to see the disorders classified with words such as axonal hereditary neuropathy whose defect is on chromosome 1. His presentation sparked considerable debate.

After a coffee break, the presentations returned to a look

at cell biology. One presentation was entitled “Role of the P2X7 receptor in the altered calcium homeostasis of Schwann cells overexpressing peripheral myelin protein 22.” Needless to say, to a nonscientist, this was a particularly difficult session to understand.

Poster sessions followed on each day and offered participants the chance to question some of the younger researchers about their studies. Some wonderful conversations were sparked by the posters, including one involving Kate Gardner from the North American CMT Database, a gentleman from the Czech Republic, and myself. He had some excellent ideas about how to improve participation by our CMTA membership in the database endeavor.

Friday was a great day for the representatives from the CMTA because one of the sessions was called “Diagnostic tools and clinical scoring for CMT” and the last session was entitled “Treatment for CMT.” Presenters discussed such important topics as how CMT patients use proximal (in this case, knees and hips) compensation to make up for distal (foot

and ankle) weakness and how this affects walking endurance and how CMT can impact on the quality of life for children. (See a more detailed discussion of this presentation on page 12). There were also discussions regarding the ascorbic acid trials in the European countries and the effects of certain orthopaedic surgeries in treating peroneal weakness. The surgeon pointed out that the most important outcome of foot surgery is the correct positioning of the heel of the foot.

All in all, the three days of presentations were amazing, inspiring, intense, and very productive. Having attended a previous North American Consortium meeting, I was somewhat prepared for the magnitude of the science being advanced, but was, once again, impressed with the brilliance and the commitment of the scientists and clinicians who not only care about the research on CMT, but also care, quite deeply, about the patients who deal with the disorder every day. As a CMT community, we should be grateful that such wonderful individuals are working on “our” disorder. ✨

**The participants in the Second International Consortium gathered before dinner on the second day.**



# GIFTS WERE MADE TO THE CMTA

## IN HONOR OF

### Eric Thomas Bolling

Mrs. Laura Bolling

### Yohan Bouchard

Mr. Richard Hanzel  
 Mrs. Arla Van Almen

### Mr. and Mrs. Bryan Gonzales

Mrs. Belle Sohnen

### Patrick Livney

Mr. Scott E. Bradway  
 Dr. Weixiong Li

### Tyler Ray Lopez

Ms. Jean Moore

### Shannon Stone

Mr. Lawrence Stone

### Mr. and Mrs. Bruce Weinstein

Mr. and Mrs. Robert Bilella  
 Dr. and Mrs. Norton Bloch  
 Dr. and Mrs. Val Bloch  
 Mr. and Mrs. James Farrell

### Keith Widdop

Mr. and Mrs. Tom Oppenheimer

## IN MEMORY OF

### Betty Chow

Mr. Henry Chow

### Nicholas K. Christie

Mr. and Mrs. George Mackey and  
 Family  
 John and Sharon Vanderpool

### Eugene Cosner

Ms. Nicole Gernux  
 Dr. and Mrs. K. J. Simpson

### Joseph D'Arcangelo

Ms. Dina Cirone

### Paul Flynn

Ms. Mona Coogan  
 Mr. George Gomez  
 Mr. James Gomez  
 Ms. Margaret Leahy  
 Mr. and Mrs. Richard Maiberger  
 Seton Hall University

### Mrs. Arla M. Van Almen

Mr. and Mrs. Richard Van Almen II

### Henry Friedmann

Mr. and Mrs. Lou Gerber

### Lee R. Gardner

Mr. and Mrs. Robert Kleinman

### Gladys Jappel

Mr. and Mrs. Jeff Sharp

### James Paul Jones

Mrs. Bertha Jones  
 Ms. Eleese C. Ray  
 Ms. Linda T. Simmons

### Robert Lee Kitchens

Mrs. Juanese Calkins

### Ruth Linker

Mr. and Mrs. Gregg Linker

### Dr. Charles T. Lynch, PhD

Mr. Tom Lynch

### Audrey Mae MacDonald

Ms. Belinda A. Bradley

### Rosemary Mahan

Mr. and Mrs. Ronald Rebain  
 Ms. Patricia Sipes

### Michael Patak

Mr. and Mrs. Neil Ambach

### Teresa Pegazzani

Mrs. Anna Maria Park

### Sarah W. Robin

Ms. Leanore Cohen

### Rosalie Sanfilippo

Ms. Rosalie Laketa  
 Ms. Rose Laketa  
 The Valentine Family

### Muriel Sheppard

Mr. and Mrs. Robert Gordan  
 Mr. Richard E. Lee  
 Dr. Henry I. Levy  
 Mr. Richard G. Williams

### Annie Vater

Mrs. Elana Gruen  
 Mrs. Linda Holmes  
 Mr. and Mrs. James Lawrence, Jr.  
 Mrs. Elizabeth Smith  
 Mr. Richard C. Toland  
 Joan, Ronald, and Alan Vater

### John Bradley

Janet R. Bradley

## 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: \_\_\_\_\_

\_\_\_\_\_

## SPOTLIGHT ON SUPPORT GROUP LEADERS

## Louise Blue Givens, PhD

Lafayette, LA is my birthplace, but I have moved numerous times. I have lived in 11 states and Washington, D.C. I think moving as often as I did helped me become a leader, organizer, and people person. Of course, I did not think moving was a positive aspect at the time. It actually helped me learn to make friends more easily.

My BA in Education was earned at the University of Arkansas and the University of Southwestern Louisiana. At the University of Oregon, I received my MA in Special Education. My PhD in Teacher Education/Special Education is from the University of Illinois, Champaign-Urbana. My career has spanned 40+ years. I have taught general and special education, college classes, and adult education in public and private schools. I have been a special education Program Specialist, Program Manager, Principal, Staff Developer, and Director of Special Education, and an Educational Consultant. I have had several professional journal articles published and have presented at numerous workshops and conferences.

I am a double divorcee. From my first marriage, I have a beautiful, talented daughter who is the light of my life.

My childhood was filled with love and family support. My two sisters are nurses and my brother is a computer master.

My brother developed

CMT ten years prior to my discovering I had the disease. I was walking down a hospital corridor toward my mom's room and heard my footsteps—slap, slap, slap. I said to myself—Good Lord, I sound like my brother. That was 1997 and I was 59 years old. Little did I know what that would mean as far as affecting my lifestyle.

Along with this disease, I also have diabetes and am a breast cancer survivor. I am strong-minded, determined, persistent, positive, open-minded, have integrity, am honest, have a great sense of humor (this helps a lot with a lot of things) and a strong sense of service. I have needed all of these qualities to rise above my difficulties.

CMT has taught me much. Being a type A personality, I walk fast, talk fast, move fast. Well, one can't continue to do that with CMT. I have had to learn to slow down physically and emotionally and to deal with a situation that was not going to improve just by sheer will.

I have had CMT for 10 years now and am handling the problems as they come. I feel blessed to not have some of the more serious limitations. Currently, I do not wear braces, can still drive my car (although hand brakes are in the near future due to lack of leg strength). I use a cane when not in my house; I am able to get about my house with environmental helps (like grabbing a wall, or shelf, or chair to stand, etc.). I do the “CMT

Dance” when I stand alone (having to keep moving to avoid falling). Carrying anything more than 10 pounds takes planning. I need to be balanced on both sides to avoid falling. I have taken several “falls” but have been lucky—no serious injuries. My arms are beginning to be like my legs—little strength and agility. My hands are very weak, as the muscles between the thumb and forefinger have disappeared. I find some packaged food products frustrating! I can hardly open a bag of potato chips! Now that's tragic!

At night I think I have restless leg syndrome, but I know it is CMT. And, there are electric shock-type pains in my legs which can occur at any time. Other than that, and a bit of arthritis, I don't have continuous pain as experienced by some.

The activities that I am doing for myself that are helpful for my condition are:

- Physical therapy twice a month has helped me with balance and strength. I am noticeably stronger than prior to having this service.
- Emotional therapy once a month has been an immense help dealing with anger, sadness, loss, grief, and all those emotions that are associated with a disability.
- Chiropractic/osteopathic service once or twice a month. I find with CMT that my bones twist out-of-place, and the few times I have fallen, this service has

fixed me up better than just time itself.

- I have done quite a bit of reading about CMT. I get the CMTA Newsletter; I am a member of the CMT Association, and I joined the Heredity Neuropathy Association. These do cost money, but the benefits far outweigh the cost.
- I have done much self-talk work to change my insidious attitude about this disability. I am into the power of positive thinking and am learning to be kind to myself. And, meditation has been one of my best techniques for rising above the “fray” of CMT.
- I am trying to break into the childrens’ book and magazine market. This endeavor has given me an incentive to remain healthy both physically and mentally. It has given me a boost of energy!
- Finally, giving back through service to others has enriched my life beyond expectations. My opportunity to be the facilitator for the CMT support group in Santa Rosa, CA has been a blessing. I get to see the strength and power of the

human spirit. The people in my group are a joy. We meet four times a year. I love to write the newsletter that reminds them of our next meeting and presenter. The stories we share and the sense of community is uplifting. It is a positive experience to share with those who share some of your own difficulties. We learn from one another, and I leave each meeting feeling better than when I arrived.

If you are not currently in a support group—try it, you’ll like it—I promise you. \*

## Golf Tournaments Help Fill the CMTA’s Coffers

BY CHARLES F. HAGINS

Board Member Patrick Livney and his brother Roland held their second successful golf tournament at the Merit Club in Libertyville, Illinois on July 30, 2007. To date, this year’s tournament has raised \$235,000 for CMT research.

Twenty foursomes enjoyed a beautiful day, weatherwise, and the challenge of the very competitive Merit course.

Dr. Michael Shy, Chairman of the CMTA’s Medical Advisory Board not only played, but presented an overview of the current science and the research strategies which were presented at the Second International CMT Consortium held in Snowbird, Utah.

Alan Pappalardo, Chicago Regional Membership Director,

presented a personal view of how CMT has impacted his life at the dinner following the tournament. A service award was also presented at the dinner, to



Patrick Torchia, President and Chairman of the CMTA.

The Livneys plan on holding the tournament on an annual basis. In just two years, the tournament has raised almost \$500,000 for CMT research.

Another successful golf tournament was hosted by AFA

Protective Systems President and CMTA Board Member, Robert Kleinman. His tournament, in June, raised almost \$70,000 for the research fund.

This year’s event was held at the exclusive Meadowbrook Club, Syosset, Long Island, New York. Fifteen foursomes competed for beautiful lead crystal trophies, presented annually to the first place team.

Mr. Kleinman has hosted a golf tournament for eight years, dedicating the profits to helping to find therapies and a cure for CMT.

On behalf of the CMTA and its members, we thank the Livney brothers and Mr. and Mrs. Kleinman and all their friends and associates for making these events successful. \*

## LIVING WITH CMT

# A Visit to the Wayne State CMT Clinic, Day Two: *The Mobility Lab*

BY DANA R. SCHWERTFEGER

To those of you who expected to see this article in the May-June *CMTA Report*, my apologies. We had thought that issue was going to be smaller than usual because of Pat's illness, but when we did the first layout, we were over our usual 16 pages and something had to be cut. Since I had not yet received my DNA test results or been fitted with my new braces, I volunteered to shelve my article for an issue...or two issues, as it turns out.

Before I describe my visit to the Mobility Lab, here's a quick update on DNA and braces: The test for CMT1X, which Dr. Shy had ordered based on his physical exam and my nerve conduction study, came back negative. Dr. Shy then ordered tests for CMT1A (PMP22) and CMT1B (MPZ), the next most likely causes based on my nerve conduction studies and profile. Those results were both negative. I'm being tested for other demyelinating forms, but so far the cause of my CMT remains unknown.

Although I've never really been concerned with what type of CMT I have, it was a little unsettling to have all the results come back negative. The greater disappointment, however, is that I have been ruled out as a candidate for the ascorbic acid clinical



Clinicians use a pad to measure balance.

trial, as well as for any potential therapeutic benefit of vitamin C.

As for braces, the orthotist in Detroit had given me a prescription for Toe-Offs, but after watching me walk with them, the orthotist here said my feet were turning out slightly and that the Toe-Offs wouldn't correct that. I could see that in the mirror, so I agreed to be cast for braces made from a material called "pre-preg"—for pre-impregnated composite fiber—and he said he would also make me molded plastic AFOs at the same time.

He gave me Toe-Offs to try in the meantime, but I never wore them. The lack of correction aside, I found it very difficult to get my foot and the Toe-Off into a shoe or sneaker.

I went back for my first fitting, and didn't have much luck getting either the plastic or the composite braces into my sneakers. I normally have the brace cut so it extends only to the ball of my foot, but this orthotist said I would benefit from the energy conservation of a brace that went the full length of my foot. Well,



the full-length braces are not only bulkier, but they also put additional pressure on the callus on my sub-fifth metatarsal, right behind my little toe.

I had expressed this concern, too, but he said he would make accommodation for it. At first, the padding seemed to be adequate, but wearing the brace for any length of time, especially while standing, is painful. I'll have to discuss options with him when I go back for the second fitting.

When I finally get my new braces in working order, it will be interesting to see if they make any difference in my scores when I return to the Mobility Lab at Wayne State next year. My visit to the mobility lab wasn't a routine part of the clinical evaluation; I just happened to be there when Dr. Allon Goldberg, a physical therapist, was

enrolling people in a study to learn how walking, balance, strength, and endurance change over time in people with CMT. The study is also gathering data on falls by people with CMT, and I was given forms to mail in every time I fall. (Fortunately, I haven't had to use one yet.)

When I arrived at the Mobility Lab in the Rehabilitation Institute of Michigan, Dr. Goldberg and his team repeated many of the strength measurements I had taken the day before, and then I participated in a variety of activities. One measured how far I could step from a standing start without losing my balance. Another measured how far I could lean forward from a standing position, and then I was asked to stand on a force plate which measured how I wobbled as I attempted to stand still with my

eyes open, and then with them closed.

I did fairly well with my eyes open, but as soon as I closed them I really wobbled. Then they put a sponge pad over the force plate, which made it feel like I was standing on a sofa cushion. I didn't do very well at that, either, especially when I closed my eyes!

The next activity involved walking down a carpet with sensors that measured various aspects of my gait, and then came the endurance test, which involved walking up and back a hallway as rapidly as I could for six minutes. I did very well at that, but six minutes seemed like a very long time.

The study should provide useful information regarding the relationship between muscle strength (or loss of strength) and balance. \*

## CMT1A CLINICAL TRIAL UPDATE

As readers of *The CMTA Report* are well aware, the CMTA and MDA are co-sponsoring the first large-scale multi-center trial for CMT1A in North America. Based on positive results in the CMT1A mouse model, patients are being treated with high-dose ascorbic acid (vitamin C). The first patients were evaluated in late April. We are pleased that approximately one quarter of the total number of patients have already been enrolled in the study. We are hoping to complete enrollment within the first year of the trial. We encourage families with

CMT1A to consider being part of this study. It is only by completing clinical trials that we will know whether compounds like ascorbic acid really have an effect on slowing disease progression in CMT1A. It is also only by completing clinical trials that we can develop the expertise to best treat and investigate additional patients and medications for CMT. We thank all of you for your help. We can't do this without you.

—*The CMT1A Clinical Trial Team*

(*Wayne State University, University of Rochester, and Johns Hopkins University*)

### For more information about the study and the study centers, contact:

Lisa Rowe  
Wayne State University  
Detroit, MI  
313-577-1689  
Email: lrowe@med.wayne.edu

Patty Smith  
University of Rochester  
Rochester, NY  
585-275-0581  
Email: patty\_smith@urmc.rochester.edu

Lora Clawson  
Johns Hopkins University  
Baltimore, MD  
410-614-4346  
Email: lclawson@jhmi.edu

# Thoughts on the CMTA's Accomplishments

BY ROBERT D. KLEINMAN

Perhaps the most gratifying part of being Treasurer of the CMTA has been to see first hand how our association has evolved from a mere information site into a moving force, underwriting research and providing tangible benefits to those afflicted with CMT.

Unlike any other charity devoted solely to peripheral neuropathies, the CMTA has done more than just talk about it; we have actually provided significant funding on many fronts. In fact, over the past five years the CMTA has utilized your donations to fund approximately two million dollars (\$2,000,000) worth of research throughout the world designed to understand and ultimately cure CMT.

The many worthy studies that the CMTA Board has chosen to fund include:

- The North American Data Base. This resource provides

those involved with the research of CMT the largest pool of information about CMT patients.

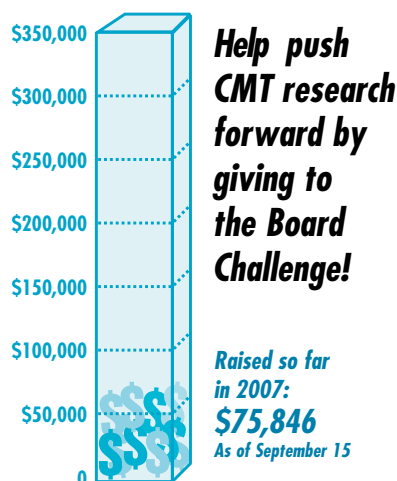
- Dr. James Lupski was awarded our first three-year grant which involves the attempt to silence the over-expression of PMP22 through various drug treatments. It is believed PMP22 mutations cause the deterioration of the myelin sheath, thereby causing some forms of CMT. Preliminary indications are that the small molecules (drugs) being used may indeed reduce toxicity and therefore may be ultimately useful as a treatment for CMT.
- Dr. Stephen Zuchner is working to determine whether mutations in Dynamin 2 could be a cause of some forms of CMT. Thus far, they have determined there is a connection between Dynamin mutations and peripheral neuropathy. They are hopeful that finding the exact molecular defect related to CMT will assist in developing drug treatments.
- Dr. Steven Scherer is examining another cause of certain forms of CMT—potassium channels. This study involves testing certain drugs to determine whether they can inhibit potassium channels in peripheral nerves and

thereby be the basis for a therapy to treat some forms of CMT.

- Ascorbic Acid Trial: During the past two years, the CMTA has been a primary supporter (donor) of this clinical trial—the first ever clinical trial specifically designed to test a potential treatment for CMT.

In addition, the CMTA is the vehicle that financially supports the mainstream medical community devoted to the study of CMT. Just last month, the CMTA underwrote the Biannual CMT International Symposium providing a forum for approximately 150 of the leading CMT doctors and scientists to meet and discuss cutting-edge discoveries and theories which will advance the world's quest to conquer CMT. We also provided funding to enable 20 young doctors, new to CMT studies, to attend the conference and thereby enlighten others in the medical community now working in this area.

Going forward we are implementing a targeted approach wherein we will underwrite projects devoted to finding the cure for CMT. The first part of this endeavor is to perfect a throughput screen. This is an important step in genetic treatment of diseases and apparently this is one area where CMT research is already ahead of others. In fact, it is very possible that our efforts here will be productive with respect not only



to CMT, but also other neurological diseases such as Lou Gehrig's disease and multiple sclerosis.

We have also devoted considerable efforts to getting government interested in our cause. Because of these efforts, for the past two years the Pennsylvania State Legislature has earmarked funding for the CMTA and its awareness program. On a broader scale, in Washington, the CMTA has worked with members of Congress to expand CMT research. As a result, for several years Congress has been urging the National Institute of Health to do so. This year, a key House Committee actually suggested that NIH capitalize on the fall 2006 workshop on peripheral neuropathies by ramping up research even more.

Finally, our financial support extends beyond just research. For example, as reported on page 2, through the efforts of CMTA, a motorized riding scooter was given to the

Over the past five years, the CMTA utilized your donations to fund approximately 2 million dollars worth of research throughout the world.

first of what we hope will be many to benefit directly from the CMTA. Being there for the presentation of the scooter to Carol Simpson with her family and the rest of the Board and

seeing the reaction first hand was indeed both emotional and gratifying. Carol, while severely affected with CMT, was so happy that day that it made us all feel like we are accomplishing something positive and that our efforts are worthwhile.

We know there are many charities out there worthy of receiving people's support. However, when considering your own financial support of an entity devoted solely to peripheral neuropathy and specifically CMT, we urge you to consider our track record. Once you do so, I am confident you will agree that the CMTA is doing the most on all fronts to tackle CMT, and you will join us in supporting the CMTA to the best of your ability. \*

## CMTA Seeks to Double Membership in 25th Anniversary Year

Next year, the CMTA turns 25. That's right. For a quarter century, the CMTA has been working hard to provide timely and reliable information to patients and physicians, to create awareness about the disorder, and to generate the resources to find a cure.

And for 25 years, you—our members—have supported these efforts, helping us to fulfill our mission and enabling us to do so much more than just publish a newsletter six times a year.

In 2008, we'll look back at the milestones we have passed and celebrate those accomplishments with you. We'll also look ahead to the challenges that still lie before us and the resources that will be required to meet them.

We've often said, "You are the CMTA!" And in that simple truth lies another: To grow, to increase our strength, to generate those resources, we must increase our membership. In fact, our goal for 2008 is to double our membership.

To accomplish that goal, we will be offering incentives to attract new members as well as to reward current members for their continuing support. And we've already started!

Between now and the end of 2007, we will give new members and renewing members\* a free CD copy of our new Patients' Guide to Charcot-Marie-Tooth Disorders.

This is fact-filled resource for both patients and physicians, and it contains features not available in the print version, so join or renew today and take advantage of this valuable offer! \*

\* Current members who renew during this period will have their membership extended one full year from their expiration date.

# Quality of Life for Children with CMT

BY PAT DREIBELBIS

One of the most interesting and, perhaps, least studied aspects of CMT in this country, is the concept that CMT can affect life in more than just the obvious physical ways. A presenter at the Second International CMT Consortium, Joshua Burns, PhD from The Children's Hospital at Westmead/the University of Sydney, Australia, discussed "The Impact of Charcot-Marie-Tooth Disease on Quality of Life in Children."

The purpose of the study was to evaluate the impact of CMT on the quality of life in children aged 5 to 18 years com-

pared with Australian age-matched control subjects. The study looked at 60 children (30 boys and 30 girls with a mean age of 11 years) whose parents completed the well-validated Child Health Questionnaire (CHQ). The CHQ asks 50 questions relating to both physical and mental health domains, including: physical functioning, emotional impact on school and friendships, physical impact on school and friendships, bodily pain, behavior, mental health, self esteem, general health, emotional impact on parent, time impact on parent, family activities, and family cohesion. Sample

questions include:

During the past 4 weeks, how much bodily pain or discomfort has your child had?

Compared with other children your child's age, in general would you say his/her behavior is—excellent, very good, good, fair, poor?

During the past 4 weeks, how much emotional worry or concern did each of the following cause you—your child's physical health, your child's emotional well-being, your child's learning ability?

Compared with one year ago, how would you rate your child's health?

## The Bugs That Live Within Me

BY HEATHER MACMICHAEL

Okay, so I'm really tired, and I'm sure that doesn't help, but I thought it might be a boredom-killer to explain the bugs that live on and in my body, and it's just my experience, so you can believe it or not.

I don't mean that I hallucinate about insects crawling on me, and I don't mean the bazillion trillion whatever-illion microscopic bugs that apparently live on and around all of us. I will also clarify that I don't mean the FBI type of bugs either.

I'm talking about the feelings I have on a constant daily

basis, that I'm SO used to that it's not really worth complaining about. Empathize with me here, nothing more, nothing less. Wear the shoes, so to speak.

And so, pardon me if I come off as crazy, but here goes.

### 1. THE ANTS

I feel ants running up and down my legs and arms all the time. I look and there is nothing there. What is actually happening because of my condition is that my neurons are sending messages, but there is a problem in the transporting of the messages, so there are false cues, which trigger a different part of my brain (or something scientific

like that), which makes my arms and legs tingle a little. It's rather annoying to try to remember not to be paranoid about it.

### 2. THE WASPS

These are the ones I complain about. They SUCK with their random stinging... wherever, whenever. Stretching doesn't make them stop; neither does un-stressing, changing my body position, diet, etc. I'm starting to suspect the weather. The wasps are unrelenting, attacking at all hours of the day and night. Sometimes they get on my last nerve (pardon the pun) and I have a mini breakdown. I am

*(continued on page 19)*

Compared with 5,414 healthy Australian children (mean age: 11.6 years), children with CMT scored lower in all areas except behavior and family cohesion. Scores were, not surprisingly, substantially lower for children with CMT compared with normal children for physical function (-29%) and its effect on school and friendships (-27%), bodily pain (-24%) and emotional impact on parents (-21%). Factors that negatively influenced quality of life in children with CMT were advancing age (physical function and its effect on school and friendships, as well as self-esteem) and CMT type (Type 3: Déjérine-Sottas produced profound physical impairment). In general, there were no major differences based on gender.

Scores for self-esteem and parental time were lower in boys than girls and advancing age resulted in a reduction in physical function and self-esteem scores.

The survey was completed by the parents and not the children themselves. The majority of the children being assessed had type 1A. In general, there were no major differences based on gender.

The conclusions drawn from this study were that the impact of CMT on children's quality of life is considerable, especially in areas such as physical function and pain, which progressively worsen with age. Dr. Burns' concluded by stating that there is a need to understand what really influences quality of life in these children so that existing and new strategies of medical care can be investigated. \*

## In the News

*From THE BOSTON HERALD:*  
Amy de Silva can't open a can of soda or a milk carton. She can't type or put a barrette in her hair because of an inherited neurological disorder that has rendered her tiny hands virtually useless.



But at age 11, she boasts a raft of accomplishments that dwarf those of most of her peers—she raises money for charities, acts in school plays, donates her hair to cancer patients, and, she'll sing the national anthem at a Red Sox [team stats] game.

"I would rather have my voice than have my hands fixed. It's such a gift and such an honor. I struggle a lot, but those obstacles you can get through and it's really, really hard to have a great voice," said Amy, a sixth-grader at Burt Wood School of Performing Arts in Middleboro.

Amy has Charcot-Marie-Tooth disease, or CMT, which affects one in 3,300 Americans, and causes the deterioration of the nerves that control sensory information and muscle function in the feet, lower legs, hands and forearms. Amy's father, William, and 8-year-old brother also have CMT, but to a lesser degree than Amy.

Most people have never heard of it.

"A lot of people say, 'What? What is it?'" said Amy's mother, Catherine de Silva of North Dartmouth. "If anybody were to see how many people it

affects daily, they would be very surprised."

De Silva worries that without more research and awareness her daughter's condition will continue to worsen. That's why she and Amy are constantly trying to raise awareness of CMT.

"We hope she can combat it with her therapy and her determination," she said.

Amy's determination seems endless.

Judy DeRossi, Amy's voice teacher at DeRossi Music School in New Bedford, said Amy was so good that she accepted her at age 6, even though she typically tells students to wait until age 9.

"She really is special," she said. "She's young. I can only expect greatness from her."

*From THE DAILY NEWS-RECORD in Virginia*

Ten-year-old Caleb Arbaugh of Mount Jackson, Virginia, has been named this year's Muscular Dystrophy Association Goodwill Ambassador for Virginia.

He appeared on the live broadcast from Gallaudet University in Washington, DC.

Caleb was diagnosed with CMT at the age of three and has traveled throughout Virginia and into neighboring states informing people about muscular disorders. He and his mother, Monica Rudy, have described their work so far as "fun and wild." \*



## SUPPORT GROUP NEWS

### Arizona – Phoenix Area

The next meeting will be held on October 25, 2007, from 6:30 PM to 8:30 PM. It will be held in the MDA conference room at 10233 South 51st Street, Phoenix, AZ. For more information, contact Aisha Hackett at 480-496-4530.

### California – San Francisco

The next meeting will be held on Saturday, October 6, 2007 at the Menlo Park Library from 2 to 4 PM. A representative from Athena Diagnostics will be on hand to show a slide presenta-

tion about the general aspects of genetic testing, the costs of the tests, and the specifics of newly available tests. Athena is the only commercial testing laboratory in the U.S. doing DNA testing for the various forms of CMT.

### Mississippi/Louisiana

A group of fifteen met on June 9, 2007, and heard a presentation by Dr. Dwight Waddell, currently the Assistant Professor of Exercise Science and Neuro-motor Control at the University of Mississippi. Dr. Waddell has a background in such disorders as

MS and Parkinson's disease. He studies movement, the effects of heat and cold and body weight as related to balance. He encouraged the attendees to control their weight and make their homes safer since those two things will improve our function as related to gait and balance problems.

The next scheduled meeting dates are September 8 and December 8, 2007.

The support group made a collective contribution of \$100 in support of the CMTA's Board Challenge.

## Help is on the Way!

Three more CMTA support groups are meeting for the first time this fall.

### Illinois – Chicago Area

The first meeting will be held on October 13, 2007, from 2 to 4 PM at the Peace Lutheran Church, Butterfield Road, Lombard, IL. This group will be facilitated by the CMTA's Chicago representative, Alan Pappalardo. If you have any questions regarding this group, please call Alan at 1-800-606-2682, extension 106.

### Michigan – Detroit Area

This group will meet for the first time on November 10, 2007, from 2 to 4 PM in the University Health Center on the lobby level of UHC (University Health Center), the

Crockett Conference Room A. This is located at the Wayne State University School of Medicine, home of the CMT clinic.

The organizer of this group is Lainie Phillips, who can be reached at 248-890-1529 or by email at familiaphillips@sbcglobal.net. The meetings will be held quarterly.

### Virginia – Harrisonburg

The initial meeting of this group will be held at the Sunnyside Room of the Sunnyside Community Center in Harrisonburg, Virginia on October 13, from 2 to 4 PM. For more information, contact Ann Long, 540-568-8328. A second meeting of the group is already scheduled for December 8, 2007 at the same location and time. \*

### Philadelphia Area

Seventeen people attended the August 18th "picnic" which was held in the community room at 2700 Chestnut Parkway, Chester, PA, the site of the national office of the association. The decision was made to have an indoor social gathering rather than risk the usual August heat and humidity in suburban Philadelphia. Of course, as luck would have it, the day was the most perfect of the summer with cool breezes and a high in the seventies. Nevertheless, the attendees enjoyed a potluck luncheon with good food and great fellowship.

The group plans to meet again on October 20, 2007, to hear Dr. Michael Shy give an update on the ascorbic acid clinical trials and other issues facing CMT patients and their families. This is a "don't miss" presentation by the Chairman of the CMTA's Medical Advisory Board. \*

# CMT Support Groups

*Support Group Liaison:* Elizabeth Ouellette, 650-559-0123

## Alabama—Birmingham

**Place:** Lakeshore Foundation  
Fitness Center

**Meeting:** Call for schedule

**Contact:** Dice Lineberry, 205-870-4755

**Email:** dklrl@yahoo.com

## Arizona—Phoenix Area

**Place:** MDA Office, S. 51st St, Phoenix

**Meeting:** Bi-monthly, Thursday  
6:30-8:30 PM

**Contact:** Marilyn Hardy or

Aisha Hackett, 480-496-4530

## California—Northern Coast Counties (Marin, Mendocino, Solano, Sonoma)

**Place:** Sutter Medical Center of  
Santa Rosa

**Meeting:** Quarterly, Saturday, 1 PM

**Contact:** Louise Givens,  
707-539-2163

**Email:** lbgivens@ix.netcom.com

## California—San Francisco Bay Area/Santa Clara County

**Place:** Location to be determined

**Meeting:** Bimonthly

**Contact:** Elizabeth Ouellette,  
650-248-3409 (C)  
650-559-0123 (H)

**Email:** elizabetho@pacbell.net

## Colorado—Broomfield

**Place:** First National Bank

**Meeting:** Bi-monthly on the  
fourth Saturday

**Contact:** Diane Covington  
303-635-0229

**Email:** dmcovington@msn.com

## Florida—Tampa Bay Area

**Place:** St. Anthony's Hospital,  
St. Petersburg, FL

**Meeting:** 2nd Sat of Feb, May, Aug, Nov

**Contact:** Lori Rath, 727-784-7455

**Email:** rathhouse1@verizon.net

## Illinois—Chicago Area

**Place:** Peace Lutheran Church,  
Lombard, IL

**Meeting:** Quarterly

**Contact:** Alan Pappalardo,  
800-606-2682, ext. 106

**Email:** alan@charcot-marie-tooth.org

## Kentucky/Southern Indiana/ Southern Ohio

**Place:** Lexington Public Library,  
Northside Branch

**Meeting:** Quarterly

**Contact:** Martha Hall, 502-695-3338

**Email:** marteye@mis.net

## Michigan—Detroit Area

**Place:** University Health Center, Wayne  
State University School of Medicine

**Meeting:** Call for schedule

**Contact:** Lainie Phillips, 248-890-1529

**Email:** familiaphillips@sbcglobal.net

## Minnesota—Benson

**Place:** St. Mark's Lutheran Church

**Meeting:** Occasionally

**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

## Nevada—Las Vegas

**Place:** West Charleston Library,  
6301 West Charleston Blvd.

**Meeting:** Email for dates 1-3 PM

**Contact:** Mary Fatzinger

**Email:** cmt\_suppgrp\_lvnnv@yahoo.com

## 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, 201-224-5795.  
Eileen Spell, 201-447-2183

**Email:** cranomat@frontiernet.net  
espell@optonline.net

## North Carolina—Triangle Area (Raleigh, Durham, Chapel Hill)

**Place:** Various locations in Raleigh

**Meeting:** Quarterly

**Contact:** Susan Salzberg,  
919-967-3118 (afternoons)

**Email:** judae@bellsouth.net

## Ohio—Greenville

**Place:** Various locations.

**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

## Pennsylvania—Johnstown Area

**Place:** John P. Murtha Neuroscience  
Center

**Meeting:** Bimonthly

**Contacts:** 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:** CMTA Office,  
2700 Chestnut St., Chester, PA

**Meeting:** Bi-monthly

**Contact:** Pat or Dana, 800-606-2682

**Email:** info@charcot-marie-tooth.org

## Virginia—Harrisonburg

**Place:** Sunnyside Community Center

**Meeting:** Quarterly

**Contact:** Ann Long, 540-568-8328

## Washington—Seattle

**Place:** U of Washington Medical  
Center, Plaza Café—  
Conference Room C

**Meeting:** Monthly, Last Saturday, 1-3 PM

**Contact:** Ruth Oskolkoff, 206-598-6300

**Email:** rosk@u.washington.edu

## "Self Care"

BY PAT MEUTH, LCSW, COUNSELOR

When we have a chronic illness such as CMT, diabetes, heart failure, or hypertension, we often have persons who care for us. We expect our doctors, physical therapists, dietitians, occupational therapists, speech therapists, and mental health counselors, to be attentive to our needs, be accurate in their prescriptions and advice, give us information when we need it, and answer our questions completely and with respect. We trust them and expect them to be available when we need them. We place our lives—our health—in their hands.

As you look at the above paragraph, do you notice someone that I have left out, someone who knows first hand your feelings and symptoms, how you respond to medications and treatments, someone who is more concerned about your health than anyone else that you

know? If you say "me," you are correct!

We are the most important persons involved in our care. Our doctors and other providers rely heavily on the information that we share with them. They, to a great extent, form their diagnosis, treatment, and prognosis based on our report to them.

"Self care" refers to a person's involvement, or engagement, in achieving the optimal degree of wellness. When we are involved in our own wellness, we consistently take responsibility for our own personal health and well-being. We "partner" with our doctors, therapists, etc.

"Partnering" includes being open to advice, listening attentively to what we are being told, following through with recommendations, taking medications as prescribed, noticing what is working and not working, and reporting this information to our

doctor or provider. Just as importantly, self care means being open and honest with ourselves.

"Partnering" also includes our support system. Do we talk to our family, friends, or someone who understands us, who can empathize with us and at the same time be truthful about how we are acting and caring for ourselves? Do we seek out help when we need it?

Achieving these goals does not happen suddenly. Self care is a process that happens step by step through life. Making small efforts toward being attentive to our own body and health and then talking to our providers is a big first step.

Dr. Michael Shy, Wayne State University School of Medicine, recently gave some excellent advice at an April 2007 CMT conference: "Be actively involved in your medical care; don't believe that nothing can be done, and educate your doctors." \*

### DISABILITY BACKLOG *(from NORD On-Line Bulletin, August 2007)*

The Social Security Administration (SSA) processes applications for Disability Insurance (SSDI) claims, and when claims are denied, it administers an appeals process that should take a few months, but often takes years. SSDI applications are supposed to take five months; but when a claim is denied, people can appeal. When an appeal is won, a beneficiary is supposed to get retroactive payments

from the month he or she initially applied. However, the SSA admits that it is currently backlogged.

About 2.5 million people file disability claims annually.

Two out of every three applicants are denied benefits.

Of those who appeal a denial, more than 60 percent eventually win benefits.

Currently, SSA has a record backlog of 745,000 pending appeals.

The wait for an appeal hearing averages 17 months.

SSDI appeals are expected to increase by 90,000 cases annually over the next five years. By 2010, there will be a backlog of one million cases each year.

Over the past six years, Congress has appropriated \$1 billion less than the President requested for administration of the SSA, according to the Social Security Commissioner. \*



*Patients' Guide to CMT...*  
**Is Now Available**

**in print...**



**and CD...**



The long-awaited update to our *Handbook for Primary Care Physicians* is now available in both print and CD format. Each chapter in the book has been written by a leading CMT expert and is directed at a more lay audience than the previous text.

Topics include the clinical features of CMT, electrodiagnostic evaluations, genetics, CMT in children, orthopaedic considerations, new treatments, physical exercise, genetic counseling, toxic medications, and resources for CMT patients and their families. The new book is 178 pages and features full color photos of many conditions and treatments.

The CD, which contains the entire text of the *Patients' Guide*, also contains a slide show by Dr. Steven Scherer, neurologist, University of Pennsylvania, which is intended for doctors who would like a quick overview of CMT and another slide show by Dana Schwertfeger, Director of Member Services, which is intended for patients and families who might need a fuller understanding of CMT.

We are currently offering the CD to anyone new who joins the CMTA before year's end or anyone who renews their membership before then.

We pride ourselves on being the best source for up-to-date information on CMT, and these two publications are our latest accomplishments in that area.

## MEMBERSHIP APPLICATION/ PUBLICATIONS ORDER FORM

(Items marked with an asterisk "\*" are required.)

\*NAME: \_\_\_\_\_ / \_\_\_\_\_ / \_\_\_\_\_  
First MI Last

\*ADDRESS: \_\_\_\_\_

\*CITY: \_\_\_\_\_ \*STATE: \_\_\_\_\_ \*ZIP: \_\_\_\_\_

\*COUNTRY/POSTAL CODE (IF NOT US): \_\_\_\_\_

\*DAYTIME PHONE: \_\_\_\_\_ EVENING PHONE: \_\_\_\_\_

EMAIL: \_\_\_\_\_ USER NAME: \_\_\_\_\_

*Required for PDF Newsletter/Website Access*

*Required for Website Access*

**Note: If you are joining now, you may purchase publications at active member prices. ACTIVE MEMBERS have paid dues within the past year. If you are unsure about your membership status, please call 1-800-606-2682.**

	QTY	COST	TOTAL
<b>ANNUAL MEMBERSHIP DUES</b>			
Members have the option of receiving <i>The CMTA Report</i> in print, PDF via email, or both.			
Receive newsletter as: <input type="checkbox"/> Print <i>or</i> <input type="checkbox"/> PDF via email		\$40	
Receive both Print <i>and</i> PDF Newsletters		\$45	
<b>NEW!</b> The Patients' Guide to Charcot-Marie-Tooth Disorders [Print Format]		<b>active members \$10</b> nonmembers \$15	
[CD Format]		<b>active members \$7.50</b> nonmembers \$10	
Charcot-Marie-Tooth Disorders: A Handbook for Primary Care Physicians		<b>active members \$15</b> nonmembers \$20	
CMT Facts I <input type="checkbox"/> English <input type="checkbox"/> Spanish		<b>active members \$3</b> nonmembers \$5	
CMT Facts II <input type="checkbox"/> English <input type="checkbox"/> Spanish		<b>active members \$5</b> nonmembers \$7	
CMT Facts III		<b>active members \$5</b> nonmembers \$7	
CMT Facts IV		<b>active members \$8</b> nonmembers \$10	
CMT Facts V		<b>active members \$12</b> nonmembers \$15	
A Guide About Genetics for the CMT Patient (No shipping and handling on this item only)		<b>active members \$4</b> nonmembers \$5	
<b>NEW!</b> Teaching Kids about CMT...A Classroom Presentation (1 hour DVD)		<b>active members \$7.50</b> nonmembers \$10	
CMT Informational Brochure and Letter to Medical Professional with Drug List		FREE	
Physician Referral List: States: _____		FREE	
<b>Contribution to CMT Research Fund</b> (100% of contribution is used to fund research)			
<b>Shipping &amp; Handling</b> (Orders under \$10, add \$1.50; orders \$10 and over, add \$4.50)			
<b>TOTAL</b>			

Check payable to the CMTA (US residents only; non-US residents, please use credit card or international money order.)

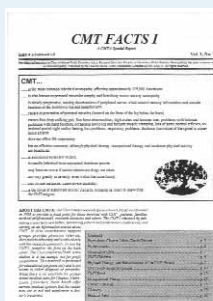
Money Order  American Express  MasterCard  VISA

Card Number: \_\_\_\_\_ Expiration Date: \_\_\_\_\_

**Mail to: CMTA, 2700 Chestnut Parkway, Chester, PA 19013; or fax to: 610-499-9267**

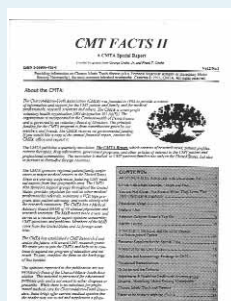
*A copy of the official registration and financial information may be obtained from the Pennsylvania Department of State by calling, toll-free within Pennsylvania, 1-800-732-0999. Registration does not imply endorsement.*

# *The CMT Facts Series:* **An Indispensable Collection of Articles about CMT**



## *CMT Facts 1*

- Facts about professionals who treat patients
- Genetics
- CMT and physical therapy
- CMT foot: surgical options
- The CMT hand
- Occupational therapy



## *CMT Facts 2*

- ADA overview
- Hope and fear
- For parents
- Anesthesia
- Adaptive gadgets
- Rehabilitative medicine
- Vocational rehabilitation
- Orthotics and orthotists
- Disability profiles
- Tremor



## *CMT Facts 3*

- Incurable, not untreatable
- CMT and pregnancy
- Prenatal testing
- Q & A about living with chronic illness
- Exercise & sports for children with CMT
- Patient services
- CMT hand surgery
- Special education
- Health insurance



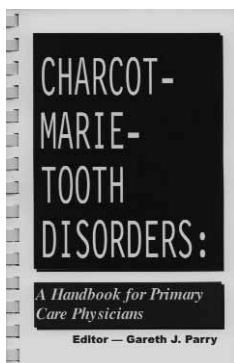
## *CMT Facts 4*

- Treatment of familial neuropathies
- Pulmonary function
- HNPP
- Orthotics survey
- Pulmonary function
- Managing pain
- Orthopedics and children
- Shriners hospitals
- Employment/ADA
- SSI



## *CMT Facts 5*

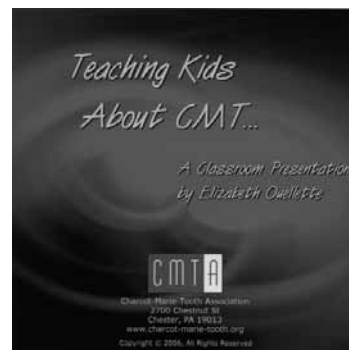
- AFOs and foot issues
- Emotional issues
- Pain and CMT
- Pregnancy
- Social Security
- Vitamins and herbs
- Physical therapy
- Occupational therapy
- Genetic testing
- Medical terminology
- Special section on HNPP



## *Handbook for Primary Care Physicians* 1995/130 pages

Edited by Dr. Gareth J. Parry, Professor of Neurology at the University of Minnesota, the *Handbook for Primary Care Physicians* is an excellent source of information about the causes, symptoms, and treatment/management of CMT. Patients will also want to read it.

**Nonmember Price: \$20.00**  
**Member Price: \$15.00**

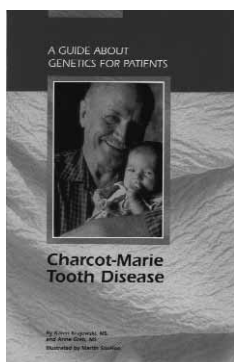


## *Teaching Kids about CMT... A Classroom Presentation*

2006/DVD 1 hr.

This hour-long DVD of an actual classroom presentation demonstrates a number of games and other exercises to teach classmates of children with CMT about the disorder.

**Nonmember Price: \$10.00**  
**Member Price: \$7.50**



## *A Guide about Genetics for Patients* 2000/21 pages

Illustrated with easy-to-understand diagrams, this booklet outlines the basics of genetic inheritance and CMT.

**Nonmember Price: \$5.00**  
**Member Price: \$4.00**


**Dear CMTA,**

I am pleased to know that the CMTA and its researchers are committed to finding a cure for CMT within the next ten years. My son-in-law and grandson both have the disease, and I am happy to support this research program for them and the many others who might benefit from it.

I hope that Charcot-Marie-Tooth disease can receive publicity throughout the country so that there is greater awareness of this disease. Hardly anyone I have spoken to in this part of the country has ever heard of it and people are surprised to learn that it is the most common inherited neuropathy. I would really like to see more publicity here in New England.

I do share in your enthusiasm for the future of CMT research, and I am looking forward to hearing from you as to the progress of that research.

—F.D.

**Dear CMTA,**

Best regards from the deep South that was battered and buried by the winds and floods of hurricane Katrina almost 2 years ago. Damage was such that everything of value belonging to my wife and me was lost to the storm. As we drove northward on the eve of Katrina's arrival with 2 cars loaded down with "important" papers, I realized I was, unfortunately, unable to bring the newsletters from CMT International. They were washed away.

During the years, from 1983-2003, I wrote a journal of episodes in my life that pertained to CMT and its effect on

me (I was a pediatrician.) for the Canadian newsletter. I had my right foot amputated and received two cardiac stents and one aortic valve replacement. During that time, I suffered complications including depression, which affected my practice. I was dependent on my co-workers, who covered for me.

This is my request: I would like to ask your readers, who might have back issues of CMT International from 1980 to look for my articles and forward a copy of them to me. My plan is to write a book and update my life in retirement.

If you can help me, you can reach me at 228-467-0347 or by email at marycayenne@bellsouth.net. Articles can be mailed to Jim Anderson, 219-B Saint Charles Street, Bay Saint Louis, MS, 39520.

—J.A.

**THE BUGS**

*(continued from page 12)*

afraid of wasps, both the real kind and the neurological sort.

**3. THE CENTIPEDES**

The centipedes are interesting. They attack mostly my legs. They are actually under my skin as opposed to on it. They wriggle around and are fairly unpleasant. Sometimes there are ants walking on top of the centipedes. They don't really bother me, but sometimes are a little frightening.

**4. THE GRASSHOPPERS**

Everyone gets the occasional twitch-type of spasm once and again, and to me, they're exem-

plified as grasshoppers. I get a LOT of these. Sometimes one spot is affected for hours. somewhere really random like one spot in my leg or my stomach. These are not painful but again, a little worrisome when they stick around for prolonged periods of time.

**5. THE BUTTERFLIES**

These are just such random tingles that happen in one spot for a moment that it feels like a butterfly is fluttering its wings right beside my skin. They almost tickle and, of all of them, are definitely the least unpleasant.

**BONUS: THE LEECHES**

Yes, I know I originally said "the

bugs," but the leeches live in there, too, so I'm going to explain them while I have your attention. The leeches are suckers, but not blood suckers. No, they latch onto my muscles, and they suck the energy right out of them. After a few minutes of walking, my legs feel like they've walked up about 10 flights of stairs. I'm not wearing heels or walking up hills. That's how the leeches are. So even when you slow down to "recharge" your energy, the leeches are still sucking. They never really stop. They suck all the time!

So that wraps up my "bug" analogy. I'll bet that more than one of you can understand exactly what I mean. ✨

**The CMTA Report** is published by the Charcot-Marie-Tooth Association, a registered non-profit 501(C)(3) health organization. © 2007, The CMTA. All rights reserved under International and Pan American Copyright conventions. No part of this newsletter may be reproduced in any form or by any electronic or mechanical means, including information storage and retrieval systems, without permission in writing from the publisher. The opinions expressed in the newsletter are not necessarily those of the Charcot-Marie-Tooth Association. The material is presented for educational purposes only and is not meant to diagnose or prescribe. While there is no substitute for professional medical care for CMT disorders, these briefs offer current medical opinion that the reader may use to aid and supplement a doctor's treatment.



## 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  
Leflunamide (Arava)  
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)  
Gemcitabine  
Griseofulvin  
Hexamethylmelamine  
Hydralazine  
Ifosfamide  
Infliximab  
Isoniazid (INH)  
Lansoprazole (Prevacid)  
Mefloquine  
Omeprazole (Prilosec)  
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  
Fluoroquinolones  
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.
- ◆ is sometimes surgically treated.
- ◆ 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 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), 1E, 1F, 1X, 2A, 2E, 2I, 2J, 2K, 4A, 4E, 4F, HNPP, CHN and DSN 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



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

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