



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 BOARD

Patrick A. Livney
Chairman and President

Gary Gasper
Treasurer

Phyllis Sanders
Secretary

Robert Kleinman
Steve O'Donnell

Elizabeth Ouellette
Jason Steinbaum

Patrick Torchia
Michael Shy, MD

Chairman,
Medical Advisory Board

Robert E. Lovelace, MD
Chairman Emeritus,
Medical Advisory Board

Charles F. Hagins
Executive Director

EMAIL CMTA AT:

info@charcot-marie-tooth.org

Celebrating 25 Years of Service and Commitment to the CMT Community

CMTA to Offer Free Membership During Anniversary Year

Beginning on May 1, 2008, the CMTA will celebrate 25 years of providing reliable information to patients and physicians, creating awareness about the disorder, and funding research at an ever-increasing rate.

This is really your anniversary and your celebration, however, because you—our members—have made the work of the CMTA possible. In addition to your membership dues, which largely support publication of *The CMTA Report* and other client services, your generosity has also made it possible for us to fund nearly \$2 million in CMT research.

We are very grateful for your support, and in recognition of all you've done for us, we will automatically renew your membership if it expires between May 1, 2008 and April 30, 2009. Instead, of the usual renewal notice, you'll receive a letter announcing the beginning of your free year.

If you are not currently a member, you will be able to join the CMTA for free between May 1, 2008 and April 30, 2009.

If your membership contributions (currently averaging

\$90,000/year) are so important to the CMTA, why are we giving them up for an entire year? The answer is that there is strength in numbers.

As we undertake more ambitious research initiatives (see center full-color section), we will be approaching major corporations and foundations for assistance. When they consider awarding grants and gifts, they look for evidence that our work is significant and that we serve a large constituency.

In this case, we believe we can better serve you by foregoing your dues for a year and using the influence we will gain as our numbers increase to seek funding from these sources.

To make this strategy work, we need each of you to encourage your brothers, sisters, mothers, fathers, or cousins—anyone

who isn't a member—to join the CMTA during this free year. They will receive six issues of *The CMTA Report*, "member" pricing on any publications they purchase, and website access.

We'll publish the form for free membership in our next newsletter, but people will also be able to join by signing up through their support group, visiting www.cmtausa.org/join (on or after May 1), or by calling 1-800-606-2682.

We are proud of everything the CMTA has accomplished in twenty-five years, and we are even more excited about what we CAN accomplish in the coming years. This is a great opportunity for everyone to be part of the effort to change the face of CMT forever. Remember: "You are the CMTA!"

—Dana Schwertfeger

INSIDE:

Devices That Help	4
CMT Triathlete	6
University of Miami Research.....	8
Support Group Spotlight	16



STAR: Changing the face of CMT Research
See page 9

"CIRCLE OF FRIENDS"

Grace's Courage Crusade

Wow! What an amazing response! Grace's Courage Crusade has started off strong and is showing all signs of continuing at a steady pace thanks to all who have been so caring and generous with their resources.

Four years ago, our daughter Grace was diagnosed with CMT and, in turn, the mystery of my own severely debilitating

disease was solved. Since there were no traces of the disease anywhere in my family and spontaneous occurrences had not been discovered until recently, I was never officially diagnosed, until Grace was.

We were under the impression that whatever my condition was it was not genetic and could not be passed on, so it was devastating when Grace was diagnosed with the same

disease as mine, which we then discovered must be CMT. After the initial shock and grief, I decided that things could and would be different for her than they were for me. I would do anything in my power to access any resource available to help ensure that she remain as physically, mentally, emotionally, and spiritually strong as possible throughout the many challenges that were to come. I also

The CMTA Circle of Friends

The people who have become involved in Grace's Courage Crusade are almost too numerous to mention, but we owe all of them our gratitude and heartfelt thanks.

Not only have Marybeth, Chris, Grace, and their family and Circle of Friends made an important contribution that will benefit all of us as we work to find a cure for CMT, they have also shown that our goal of raising \$10 million for research is within reach!

Granted, we can't expect every Circle to be as successful, but we don't need it to be. If you start a Circle of Friends and raise \$5,000, we'll only need 1,999 other people to do the same and we'll have that \$10,000,000 in no time at all.

Do you believe that's possible? We do, and we'll do whatever it takes to help you with your Circle of Friends. But we can't do it unless you're willing to help, so please call us today at 1-800-606-2682, email us at cof@charcot-marie-tooth.org, or visit us on the web at www.charcot-marie-tooth.org/cof and get started!

A B Hoxie, Inc.
Mr. and Mrs. Alfred R. Abbatiello
Access Communications, LLC
Mr. and Mrs. Hisham Aharon
Mr. and Mrs. George Ainsworth
Drs. Joseph and Seena Aisner
Albert Ranallo Plumbing & Heating
Mr. and Mrs. Lincoln D. Almond
American Endowment Foundation
Mr. and Mrs. Gary W. Anderson
Mr. and Mrs. John Anderson
Mr. Mark Andrews
Ms. Emelie Arruda
Art In Motion Dance Company
Mr. and Mrs. John V. Artale
Mr. Salvatore J. Augeri
Mr. and Mrs. Nicodemuz Ayala
Mrs. Carolyn S. Bach
Mr. and Mrs. Charles L. Bach, Jr.
Ms. Caryl Lee Bank
Mr. and Mrs. James Barbera
Mr. and Mrs. Eric Barboriak
Mr. and Mrs. Stephen Barbosa
Mr. and Mrs. Bruce R. Barrett
Mrs. Angela Batel
Mr. and Mrs. Glen C. Beauchemin
Mr. and Mrs. Charles Beck
Dr. Peter J. Bellafiore
Dr. Gerald J. Berrafati
Mr. and Mrs. Kevin and Josephine Berrafati-Tice
Mr. and Mrs. Edward J. Bianchi
Mr. and Mrs. Irwin M. Birnbaum
Mr. and Mrs. Norm and Marcia Blackburn
Mr. and Mrs. Charles L. Blanchette
Mr. and Mrs. Louis D. Boccardi
Mr. and Mrs. David and Gloria M. Boylan
Mr. and Mrs. James Brauer
Mr. Gregory R. Brazil
Mr. and Mrs. Joseph Brian
Mr. Arthur Briggs
Drs. Dante and Nance Brittis
Ms. Jacqueline Brocco
Dr. and Mrs. James and Ann Brown, Jr.
Mr. Robert Caffrey
Ms. Alberta R. Caldarone
Mr. and Mrs. Paul Cantatore
Mr. and Mrs. Ronald L. Caouette, Jr.

Mrs. Carol Capalbo
Mrs. Jane Carlson-Pickering
Ms. Diane M. Caroselli
Ms. Carole A. Carroll
Mr. and Mrs. Peter Carton
Mrs. Cynthia Castellone
Dr. and Mrs. Anthony Catuogno
Ms. Janice Causey
Mr. and Mrs. Thomas F. Cerussi
Mr. and Mrs. Robert Chabot
Chariho Middle School Staff
Chariho High School PTO
Mr. and Mrs. Robert R. Chartrand, Jr.
Mr. and Mrs. Francis D. Cherenzia
Ms. Viola M. Chorney
Dr. and Mrs. Vincent Ciaramella
Ms. Linda D. Ciolfi and Ms. Ros Johnson
Mr. and Mrs. John T. Clark
Mr. and Mrs. Constantine Clemente
Mr. and Mrs. Joseph and Sharon Clossick
Mrs. Lori Ann Sidoti Clymas
Ms. Arlene M. Coccia
Mr. and Mrs. Bernard C. Coghlan
Ms. Lorraine Colasuonno
Ms. Linda C. Cole
Mr. and Mrs. Robert Connell
Mr. and Mrs. Christopher Connery
Mr. and Mrs. Michael Cooperman
Mr. and Mrs. Thomas B. Coppola
The Corcoran Family
Corporate Laser, Inc.
Mr. and Mrs. Steven Corvese
Mr. and Mrs. Arthur and Lorene Cripps, Jr.
Mr. Nicholas Croce
Mr. and Mrs. Stephen and Veronica Cuhna
Mr. and Mrs. Frank Curcio
Mr. and Mrs. Dean and Julie D'Andrea
Drs. Domenick and Valarie D'Angelica
Dr. Thomas A. D'Eletto
Mrs. Susan C. Davis
Mr. and Mrs. Alex de Borja
Ms. Enes De Carlo
Mrs. Theresa De Divitis
Dr. and Mrs. Anthony De Martino
Mr. and Mrs. George and Carol M. De Roeck
Mrs. Evelyn Deletto
Mr. and Mrs. Kevin J. DeLoge, Sr.

Mr. & Mrs. David DeLorenzi
Ms. Anne DeLorenzo
Ms. Donna DeLorenzo
Mrs. Maria C. DeLorenzo
Mr. Christopher P. DePhillips
Mrs. Jeanne A. Deus
Mrs. Linda J. Di Gilio
Mr. and Mrs. Rob DiCocco
Mr. and Mrs. Scott and Helen Douglas
Mr. and Mrs. Bruce D. Downer
Mr. and Mrs. Leonel J. DuBois
Mr. and Mrs. James K. Duff
Mr. and Mrs. Phillip J. Duffy
Mrs. Rosemary Dumberger
Mr. and Mrs. Patrick C. Dunican, Jr.
Mr. and Mrs. John J. Dunleavy
Mr. and Mrs. Paul M. Dunn
Ms. Cheryl Dyer
Ms. Joan C. Egan
Jim, Kalen, and Jess Elliot
Mr. and Mrs. James K. Engel
Mr. and Mrs. Richard Esposito
Ms. Alice Esposito-Hunt
Eufemia Jewelry
Mr. Randy Even
Mr. and Mrs. Kenneth and Andrainna Falcone
Ms. Barbara Feingold
Mrs. Jaclyn M. Ferria
Mr. Howard K. Fielding, Jr.
Ms. Dawn B. Fishman
Mr. and Mrs. Richard Fiske
Mrs. Victoria Foster
Mr. and Mrs. Leo H. Fox
Mr. and Mrs. William F. Gallivan
Mr. and Mrs. Frank J. Gallucci
Mr. and Mrs. Preston S. Gardiner
Mr. Jeffrey Gardner
Mr. and Mrs. R. Scott Garley
Mr. and Mrs. John M. Gennari
Mrs. John B. Gest
Mr. and Mrs. John Ghiorzi
Mr. and Mrs. Edward Giannetta, Sr.
Mr. and Mrs. Matthew Giannetta Gibbons, P.C.
Mrs. Assunta Giglio
Mr. and Mrs. Tom and Lorie Gilligan
Mr. and Mrs. Joe Godino



Grace and her mother Marybeth have had great success with their Circle of Friends.

decided, after our first visit to Dr. Shy and his team out at

Wayne State and several conversations with Elizabeth Ouellette and Dana Schwertfeger, that it would be essential to participate in the efforts to help advance research as quickly and effectively as possible.

The Circle of Friends seemed the way to go, and four months and \$60,000 later we are thrilled to report that we feel truly blessed! The unending support of family, friends, coworkers, fellow parishioners, neighbors, teachers, and outright strangers who were touched by our story, cared, and contributed has been overwhelming!

Not only have we been showered with financial generosity, but the thoughts, kind words, prayers, and efforts of others have been invaluable and continue to inspire us as we forge ahead. Thanks to many of them, we look forward to upcoming school fundraisers, a spring bike-a-thon, and a summer dinner dance! We are encouraged to see the beginnings of research breakthroughs and know together, in the name of Grace's Courage Crusade, we can help change the futures of those living with CMT.

—Marybeth Caldarone

Mr. and Mrs. Richard Goldberg
Mr. and Mrs. Michael Gordon
Mr. Keith Gotti
Mr. and Mrs. Ron Gramaglia
Dr. and Mrs. Luke F. Grande
Mrs. Rodna L. Grande
Dr. and Mrs. Peter and Ruth Grant
Hairwaves Salon
Mr. and Mrs. John Hamill
Mr. and Mrs. Robert S. Hanley
Ms. Kathleen J. Harrington
Mr. and Mrs. Robert Harrington
Mr. and Mrs. Richard N. Harris
Mr. and Mrs. Russell R. Harris
Mr. and Mrs. Lewis Hassell
Ms. Carol A. Hathaway
Mr. and Mrs. Owen Haynes
Mrs. Julie Ann Heim
Mr. and Mrs. Gerard Heller
Ms. Florence Henderson
Mrs. Bonney B. Henschel
Mr. and Mrs. Robert P. Hildum
Hogan & Hogan, Ltd.
Mr. and Mrs. Clifton G. Hon
Ms. Tina M. Hoover
Mr. and Mrs. Francis X. Hopkins
Mr. and Mrs. Edward D. Hutchinson
Mrs. Barbara L. Irmischer
Jewish Communal Fund
JPMorgan Chase Foundation
Ms. Vivian D. Julier
Mr. and Mrs. George and Judith Karam
Mr. and Mrs. Glenn Kendzia
Ms. M. Thelma Kenyon
Mr. and Mrs. John F. Killoy, Jr.
Mr. and Mrs. Don and Eileen Klein
Mr. and Mrs. John Klock
Mr. Kevin Kramer
Mr. and Mrs. Richard J. La Magna
Mr. and Mrs. Anthony W. Lachowicz
Ms. Mariellen C. Langworthy
Ms. Dyana Lee
Mrs. Phyllis Lefoley
Mrs. Mary E. Lenihan
Mrs. Leslie Liebman
Mr. and Mrs. Robert Lindeblad
Mr. and Mrs. Carol Liu
Mr. Chris Livingston

Mr. and Mrs. Joseph P. Lombardi
Mrs. Marian A. Lombardi
Ms. Nancy Lottinville
Mr. and Mrs. Michael and
Waltraud Lukelitch
Mr. and Mrs. William Lupinacci
Mr. and Mrs. David M. MacDonald
Mr. and Mrs. John D. MacDonald
Mrs. Dominique Machado
Mr. and Mrs. Richard Macksoud
Mr. Kevin Mahoney
Mr. and Mrs. Anthony Marino
Mr. and Mrs. Irving Martin
Mr. Timothy J. McHugh
Mr. and Mrs. Christopher M. McLoughlin
Mr. and Mrs. Gerard McLoughlin
Mr. and Mrs. Peter McLoughlin
Mr. and Mrs. Brian McMahon
Mr. and Mrs. Frank W. McNally
Mr. and Mrs. Robert C. McVey
Ms. Brenda Medina
Ms. Rosemarie Medina
Mr. and Mrs. John W. Miller, Jr.
Mrs. Angelina Mincin
Dr. and Mrs. Jules E. Moneagle
Mrs. Kathryn Montague-Magnotta
Mr. and Mrs. William Moran
Mrs. Nancy Moreira
Mrs. Catherine A. Moskosky
Mr. Debashish Mukhopadhyay
Mr. Terry Myers
Mr. and Mrs. Vito J. Navarro
Ms. Patricia Negavrab
Mr. and Mrs. Chad W. Neighbors
Mr. and Mrs. Lane W. Newquist
Ms. Diane M. Nydick
Mr. and Mrs. Michael O'Byrne
Mr. and Mrs. James O'Donnell
Mrs. Mary O'Hea
Ms. Jean G. O'Malley
Mrs. Kate O'Malley
Mr. and Mrs. Joe and Katherine O'Reilly
Ms. Elizabeth Odierna
Mr. and Mrs. Christopher J. Orefice
Mr. and Mrs. Kenneth R. Orefice
Mr. Stuart Orefice
Mrs. Joan M. Ott
Ms. Marianne Papi-Roque

Mr. and Mrs. Carl A. Papino
Ms. Tonia C. Papitto
Mr. and Mrs. Robert M. Parrillo
Mr. and Mrs. Frank Patnoad
Mr. Henry S. Patridge, MD
Mrs. Laurie Pearce
Mr. and Mrs. Frank A. Pensa
Mrs. Jennifer Pepperman
Mr. and Mrs. John Perrette
Dr. and Mrs. Gordon Peterson
Mrs. Kristen Petit
Pfizer Inc.
Steven M. Pinch, LLC
Mr. and Mrs. Peter A. Piscitelli
Ms. Geraldine Ponto
Ms. Elizabeth Procaccianti
Ms. Eve Procaccianti
Procaccianti Group
Mr. and Mrs. Mark R. Provost
Mrs. Alice E. Quinn
Mr. and Mrs. Michael Quinn
Ms. Susan L. Quinn
Mr. and Mrs. Anton Radovic
Mr. and Mrs. Frank Reilly
Ms. Cynthia Reiser
Mr. and Mrs. Keith Renner
Mrs. Dolores Rhodes
Ms. Dora A. Rhodes
Ms. Dolores Rivellino
Mr. and Mrs. Eugene Rizzo
Mrs. Maria T. Rodriguez
Mrs. Susan T. Rogers
Ms. Fern G. Ruhalter
Ms. Barbara Ann Russo
Mr. and Mrs. Everett Salisbury
Santa Claus
Mr. and Mrs. Barry Scott
Mrs. Nikki G. Scott
Mr. and Mrs. Chris Senard
Mr. and Mrs. S. T. Sharkey
Mr. and Mrs. Shawver
Mrs. Nancy W. Sherman
Mr. Steven Sholk
Dr. and Mrs. Robert Shprintzen
Dr. and Mrs. Eugene J. Sidoti
Mr. and Mrs. Sal and Rosalie Sidoti
Mr. and Mrs. Jeffrey B. Sincoski
Mr. and Mrs. Richard J. Siravo

Sisters of St. Francis
Dr. and Mrs. Peter Small
Mr. and Mrs. Edward A. Smith
Mr. and Mrs. Matthew O. Smith
Mr. and Mrs. Robert and
Elizabeth M. Smith
Rev. John P. Soares
Mr. and Mrs. Ted and Karen Sorlien
Mr. and Mrs. Joseph Sparacio
Mr. and Mrs. Jeffrey Sparfven
Dr. and Mrs. George A. Spreccace
Ms. Jill St. Andre
St. Pius X. School
Ms. Ann E. Steidle
Mr. and Mrs. Michael G. Steinberg
Mr. and Mrs. Ross A. Still
Mr. and Mrs. Timothy J. Sullivan
Ms. Maria Sweet
Mrs. William Synan
Mrs. Sarah Ann Szumita
Mr. and Mrs. Joe Thompson
Mr. and Mrs. Joseph Torrente
Mr. and Mrs. Peter A. Torrente
Mr. and Mrs. George H. Travis
Mr. and Mrs. Philip J. Tulotta
Ms. Mary Turrissi
Ms. Mary Ellen K. Twomey
United Way of RI
Mr. and Mrs. Vincent Vetrini
Dr. and Mrs. Paul Vilmos
Mr. and Mrs. Alan W. Walker
Mr. and Mrs. James D. Warburton
Mr. and Mrs. Jason H. Ward
Mr. John J. Watras
Mr. and Mrs. Michael Webster
Mr. and Mrs. Robert J. Webster
Mr. and Mrs. Robert Webster
Wyeth
Mrs. Barbara A. Zaborski
Mrs. Etta Zasloff
Ms. Mara E. Zazzali-Hogan
Dr. and Mrs. Lester Zimmerman
Mrs. Deborah A. Zonfrilli

Helpful Devices for CMT Patients

BY ALAN PAPPALARDO

My name is Alan Pappalardo and I have been dealing with the effects of CMT 2A for over two decades. In the course of my daily routine certain things have proven useful in overcoming CMT. The following three reviews pick items ranging from the simple to the highly sophisticated; however, each helps the day go by as effortlessly as possible.

HEMOSTATIC CLAMPS FOR INCREASED DEXTERITY

If you have ever watched a medical show, maybe *M*A*S*H* or *ER*, you are sure to have seen the doctors use small metal clamps in their practice. These medical tools are actually called hemostatic clamps and are a cheap and effective way to increase your dexterity. As almost every CMT patient understands, simple tasks such as picking up a coin or retrieving a straight pin that fell into a drawer can be laborious if not infuriating. Hemostats are designed to perform very precise grabbing with the minimum of effort. The clamps also have a secondary function that locks them in place which is useful in many applications like pulling



on fabric or holding papers in place. While hemostats take some practice to master, most CMT patients will be able to utilize their benefits. Hemostats are a low-cost option to help combat the daily struggles that muscle degradation brings forth. You can find a variety of hemostatic clamps at any medical supply store and at most hobby shops for around \$5-\$15. American Science and Surplus sells hemostats and other similar devices for a very reasonable price: <http://www.sciplus.com/category.cfm/subsection/12/category/128>

VOICE RECOGNITION SOFTWARE THAT RENDERS THE KEYBOARD OBSOLETE

Over the last few years voice recognition software has improved by leaps and bounds. In the past using such software could take more time than crossing the Oregon Trail and leave you so frustrated that it felt like you just finished your taxes. The title that stands out among all others is Dragon Naturally Speaking Preferred. Right out of the box, without any training Dragon can achieve 70% accuracy. With only 20 minutes of training and the reading of a small document, Dragon's accuracy increases to about 90 to 95%. One can further increase that value by manually inputting words, or even easier, by letting the program adapt through simply using its primary function. Not only does Dragon take your



voice and turn it into text, there are a multitude of other features. Verbal commands allow Dragon to interact with many common programs one uses in everyday life, like Microsoft Office and database software suites. Dragon also has a numbers mode that makes inputting data a breeze. Finally, when you have finished writing an email, or producing that important document for the boss, Dragon will read your text back to you so that you can double check for any errors. While voice recognition software is not perfect, it has evolved to the point where incorporating it into your life is a seamless process and one that you will wonder how you lived without. Dragon Naturally Speaking Preferred can be found at most computer or technology stores for about \$200. TigerDirect.com sells Dragon for \$190.

WARM AIR BLANKETS OR HEATED BLANKETS?

One problem that CMT patients face is poor circulation in the extremities: how do you keep your arms and legs warm? When ordinary blankets are not

sufficient, hospitals turn to warm air blankets. Warm air blankets are ideal for heating someone who has poor circulation. Heated air is evenly distributed throughout the blanket which avoids the problem of 'hot spots. Unfortunately, warm air blankets require a precision air pump that takes them out of the price range of the ordinary individual. An economical substitution is simply the electric blanket. The current choices are not your grandfather's heated blankets. They are safe and reliable with little chance of fire when used properly. Electric blankets use the resistance of metal in an insulated wire running throughout the product to create heat. Sometimes some sections can be warmer than



others, so caution is advised when using these blankets. However, when properly supervised and correctly operated, electric blankets are a reasonable solution for a CMT patient's cold legs. Those who are interested in purchasing warm air blankets, can visit the website for Cincinnati Sub-Zero at www.cszmedical.com. Expect to pay around \$2000; that price is standard in the industry. On the other hand, a fantastic heated blanket made by Sunbeam can be purchased for about \$50-\$75 at www.sunbeam.com or at other retail sites. *

COPING WITH DAILY TASKS

Ideas from Readers

This is a new column dedicated to readers' solutions to dealing with CMT and the ordinary tasks that are made difficult by its progression. These ideas come from Abby Wakefield of New York:

- ☆ Each time you go to a new physician, take a copy of the last page of *The CMTA Report* with you since it has a brief, but complete enough explanation of CMT, the toxicity of certain drugs, the person's name who received the newsletter, and it would be a good idea to date that page to document the time frame of the information. I have been doing this for years, and each new physician is more than interested to receive this little summary of the disease to be put in my chart. Most physicians do not know about CMT, so to present this page of information about our disease helps them to have a permanent piece of paper to reference.
- ☆ Carry a copy of the last page of *The CMTA Report* with you in your purse or wallet in case of an emergency. Also, compile a listing of your current medications and the dosages. Any supplements you take and those dosages too should be listed. It is also important to list emergency contacts and the name of your primary care physician, and any other specialist you see on a regular basis. You can probably get all the information on one sheet of paper, and it folds nicely in order to not take up much room in your purse or wallet. If there is an emergency, that list plus a copy of the last page of the CMTA newsletter will be invaluable to people treating you.
- ☆ Carry rounded-top metal baby scissors in your purse. These scissors can be purchased at any chain drugstore. They are great for cutting open those items packaged in indestructible packaging, from potato chips to pretzels and everything in between. If you fly, before you go through security put the scissors in the plastic ziploc baggie in which you put your three-ounce liquids, rather than leaving the scissors in your luggage or purse. I have never, in all the years since 9/11, had these scissors taken away from me. They come in handy for so many items that cannot be easily ripped open by us anymore. *

Send Us Your Strategies...

If you have ideas to share with our readers, please email them to the CMTA at info@charcot-marie-tooth.org.

CMT: A Curse Turned into a Blessing

My name is Shaun Daley, and I have Charcot-Marie-Tooth. CMT is the most commonly inherited peripheral neuropathy and is found world-wide in all races and ethnic groups. CMT affects an estimated 2.6 million people.

I live near Peachtree City just below Atlanta and have a wife and two children. I am 29 years old and have a very active lifestyle. As a young boy I did not realize that I had CMT until signs started in my early teens. My ankles became very weak, and I would often trip and fall down not knowing why. I participated in high school football and did not know I had this disorder until the signs were so bad I had to get it checked out. My mother and sister, who had the same symptoms, went with me to a nerve specialist in Atlanta and that is when all three of us were diagnosed with CMT.

My life suddenly changed. What I thought was sure to be a curse has actually been the biggest blessing aside from having met my wife and having my two boys. Soon after high school, I found that my days of running were over because of the onset of the disorder's ugly side effects. However, I did not let this stop me, so I started rac-

ing motorcycles. I found this sport was the only thing I could do to stay competitive in some fashion, and I got good at it. While having a lot of success in this sport, I also encountered many injuries. I loved racing motorcycles, but that did not compensate for my not being



Shaun's ultimate goal is to compete in an Ironman Triathlon.

able to run and be competitive in other sports.

As a training measure for motorcycle racing, I would cycle with my two brothers-in-law who competed in the sport of triathlons. I admired them for what they did, and they really encouraged me to get involved in the sport. I thought to myself: I know I can swim, and I know I can bike, but I can't run. I studied the anatomy and the

biomechanics of the ankle and did a lot of research on possible orthotics that could provide me with the support and functionality required to not just improve my walking, but actually allow me to run. After this search ended in a dead end, I began to search for an athlete with this disorder who might have experience with orthotics or shoes that would make jogging possible. This also ended in a dead end.

Even though I ran into several challenges along the way, I did make some great connections with people who really care and are now supporters of my efforts like The Getting 2 Tri Foundation and the Challenged Athletes Foundation. I finally took it upon myself to go to an orthotics company and tell them what I needed. I actually helped them make the shoes I am running in now. I now have AFOs (braces) and custom shoes that mimic the gait of an able-bodied person to some extent. I had not run in 12 years! I started jogging with the aid of my shoes and braces and felt like my journey had just started. Indeed it has!

I entered a triathlon in Sept. 2007 and was named the state champion in the physically challenged category at the Peachtree City sprint championships. I am very proud of this accomplishment! So where am I going with this and why is it so important?

(continued on page 17)

Different Ways to Support the Work of the CMTA

GIFTS OF STOCK

A gift of appreciated stock can save you taxes when you make a donation to the CMTA.

To make a donation of stock or other securities (no cash or checks, please) send information to the Charcot-Marie-Tooth Association, Account # JH 45207-53, Federal Tax #22-2480896, and please contact our broker:

UBS
1735 Market St, 35th Floor
Philadelphia, PA 19103
DTC#221
Attn: Carol Thompson
Tel: (215) 972-6841
Fax: (215) 972-6801

So that we may acknowledge your donation, please send copies of your letter to Kim Magee at the CMTA, 2700 Chestnut Pkwy., Chester, PA 19013 or call Kim at (610) 499-9264 or (800) 606-2682.

You may wish to learn about other gift-giving opportunities by consulting your attorney, accountant, and/or tax or estate planner.

MAKING A BEQUEST

There are a number of ways of continuing your interest in the Charcot-Marie-Tooth Association through your estate plans. One simple and meaningful way is to include a bequest in your will. Your attorney can help prepare your will and choose the best wording for your particular circumstances. For an example, your will (or supplemental codicil, if you do not wish to write a new will) could state:

“I give and bequeath to the Charcot-Marie-Tooth Association, a not-for-profit corporation, organized under the laws of the Commonwealth of Pennsylvania, and having its principle

office at 2700 Chestnut Pkwy., Chester, PA 19013, the sum of \$() or () percent of the rest, residue, and remainder of my estate to be used for the (general purposes) or (research fund) of the Organization.”

A bequest to the CMTA is fully deductible for estate tax purposes. Additionally, you will be providing hope to CMT patients and families now and in the future. ✨



WE'RE ALMOST THERE!

Thanks to all of you who gave to the Board Challenge. We've almost reached our goal, which will help us move forward with our ambitious plans for our 25th anniversary year.

**As of March 11th, 2008:
\$348,194**

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: _____



GIFTS WERE MADE TO THE CMTA

IN HONOR OF

Yohan Bouchard

Ms. Patti Rooney

Cathy Brooks

Mr. & Mrs. Kenneth Dunham

Jack A. Graves

Mr. John R. Graves

Margie Kelly

Mr. Carl Burnham

Shannon Kelly

Mr. Shannon Kelly

Tyler Ray Lopez

Mrs. Jean A. Moore

Beverly O'Free Rooney

Ms. Patti Rooney

H. Geoffrey Sheffer

Mr. William A. Bradley

Phil & Carol Furnas

H. Geoffrey Sheffer (cont.)

B. G. Hudson

Ms. Tracey L. Kita

Mrs. Fay Rice

Dr. Michael Shy

Ms. Carolyn A. Koski

Keenon F. Werling

Mr. Silas M. Ford III

IN MEMORY OF

George G. Bledsoe

Mr. & Mrs. Frank Gunnison

John S. Bradley

Mrs. Janet S. Bradley

Shirley R. Bunis

Ms. Mary Alderman

Ms. Ruth Cohan

Ms. Anita R. Goldstein

Ms. Donna B. Kranitz

Mr. & Mrs. Abe Sitcov

Bessie Carnel

Ms. Barbara Whatley

Paul Flynn

Westchester/Fairfield County

Support Group

J. A. Foster, Jr.

Mr. John A. Foster, III

William Ray Fountain

Geralyn K. Beahm

Mr. & Mrs. Walter A. Clark

Mr. & Mrs. Richard H. Rieber

Sue Frahm

Ms. Donna Brill

Mr. & Mrs. Howard Kesner

Ms. E. Mindy Nussbaum

Nettie Friedman

Mr. & Mrs. Robert Kleinman

Shirley Friedman

Meltzer Family

Henry Friedmann

Ms. Agnes Aronsohn

Hannah Greenstein

Mr. & Mrs. Eugene Sidoti

Howard A. Parker

Ms. Patricia Davis

Mr. & Mrs. William Howard

Ms. Faye D. Richey

Rebecca Sand

Rhoda & Stephen Sand

James Smith

Ms. Susan Gesing

Mr. Anthony L. Janulewicz

Ms. Stacy B. Talbott, Esquire

Wallace Wayne Wyatt

Mr. & Mrs. Robert Colby

RESEARCH TEAM SEEKS FORMER PARTICIPANTS

In 2007, Drs. Jeffery Vance and Stephan Zuchner and members of their CMT research team moved from the Center for Human Genetics at Duke University to the University of Miami, Miller School of Medicine. They are now faculty in the newly established University of Miami Institute for Human Genomics.

CMT research will remain a major focus of these investigators at the University of Miami, including their ongoing research in CMT. In addition, this move is creating exciting new opportunities to expand their CMT research. This includes reaching more families, providing additional resources for both clinical and laboratory work on CMT, and facilitating new collaborations.

The research at Duke will close. All laboratory work and data analysis will now take place at the University of Miami Institute for Human Genomics. The University of Miami will be the center for all family and patient studies.

Drs. Vance and Zuchner and their team fully realize that without the participation of families their study could not exist or continue to thrive. The move to Miami involves transferring the existing research samples and data from Duke to the University of Miami Institute for Human Genomics. The same level of security and confidentiality exists at the University of Miami Institute for Human Genomics as did previously at Duke.

All participants were sent a letter informing them of this

change. However, the research group anticipates some participants may not receive the letter because they have moved. Therefore, to ensure that as many participants as possible are notified of this move, we are posting this information.

If you participated in this research project and do not want your samples and data used by the University of Miami, or have questions about this transition, please contact the study personnel at 1-800-385-3244, or the research team in Miami at 1-877-6UM-MIHG (1-877-686-6444).

In closing, Drs. Vance and Zuchner hope that the CMT community will find this news as exciting as they do and they look forward to continuing their work into the genetics of CMT. *

Imagine a future



without CMT...

STAR: Changing the faces of CMT

The CMTA La

A Strategy to Ac

For 25 years, the vision of the CMTA has been a world without CMT.

Now, the vision has the potential to become reality as research moves to a new level at an increased speed.

CMT is unlike other neuromuscular disorders because its causes have been pinpointed, leading to the identification of at least 33 specific gene defects. More importantly, the fact that these genetic mutations can be replicated in laboratory models and grown as tissue cultures opens an extraordinary window of opportunity to develop treatments and cures for CMT in the immediate and foreseeable future.

To accomplish these goals, the Charcot-Marie-Tooth Association (CMTA) has established the Strat-

egy to Accelerate Research (STAR)* to specifically fund CMT-related research. The scientists or Principal Investigators conducting research for the STAR initiative have been chosen from an international body of the world's most accomplished medical pioneers. The STAR program's unique character stems from the willingness of each and every one of these prominent researchers to come together to advance CMT research as a team, sharing and communicating ideas, discoveries and research findings.

BACKGROUND

CMT1A, the most common form of CMT, is caused by a duplication of a gene which causes an overproduction of a protein (PMP22). PMP22 and myelin are made by cells known as Schwann cells. Myelin is the insulation that surrounds the nerve fibers, which are also known as axons. The over-

* The STAR program is modeled after the Myelin Repair Foundation's Accelerated Research Collaboration (ARC).



“The Strategy to Accelerate Research is the most important initiative ever undertaken by the Charcot-Marie-Tooth Association. In addition to developing specific therapies for CMT patients, the translational science employed in the research could have major implications for the treatment of a host of related genetic disorders.”

—Dr. Michael E. Shy, director of the CMT Clinic and co-director of the Neuromuscular Program at Wayne State University, and chair of the CMTA Medical Advisory Board

Unleashes STAR: Accelerate Research™

expression of PMP22 causes the deterioration of the myelin sheath that surrounds the axon, ultimately resulting in the deterioration of the axon itself. As a result of this process, vital nerve impulses are no longer able to transmit efficiently, resulting in the weakness and loss of sensation that characterize CMT1A.

Two different medications (vitamin C and a progesterone antagonist, onapristone) have already been shown to reduce PMP22 levels to a more normal expression in laboratory models of CMT1A. Neither has been validated in clinical trials in humans, although trials of vitamin C are underway in several European countries and in the United States (the US trial is funded by the CMTA in partnership with the MDA). The progesterone antagonist (onapristone) used to treat CMT1A in laboratory models isn't suitable because of its toxicity in humans. Since there are surely many other potential but undiscovered compounds currently available to lower PMP22 levels and treat CMT1A, our aim is to develop a screening methodology to identify as many compounds simultaneously, in the least amount of time possible. This is feasible with currently available technology through a process known as "high-throughput screening," in which various compounds are tested using robots and cultured Schwann cells to screen tens of thousands of candidate compounds to determine whether they can lower PMP22 levels.

By partnering with pharmaceutical companies and some of the best Schwann cell scientists in the world, we will endeavor to test hundreds of thousands to millions of compounds. The "hits" generated in this screening process will be validated, through ever more stringent tests, including a laboratory model of CMT1A. The goal is to develop at least two compounds during Phase 1 trials within a 3-year period.

THIS YEAR'S THREE-PRONGED RESEARCH INITIATIVE

In order to reach our ambitious goal of developing compounds that can treat CMT1A, the CMTA is funding the following three projects:

Project 1

Development of a stabilized cell line that expresses PMP22

Using actual Schwann cells, as well as Schwann cells combined with tumor cells (which promote growth), we plan to create a cell line which researchers will use with high-throughput screening to identify which compounds currently in use by drug companies work to diminish the amount of PMP22 in a cell. Thousands of medicines can be placed in tissue culture plates. By utilizing fluorescence or luminescence, researchers are able to observe which medicines dim the fluorescent glow, suggesting that a particular compound is lowering the amount of PMP22 being produced. Since CMT1A involves the overproduction of PMP22, this procedure should quickly produce "candidate" medicines to control that overproduction.

Project 2

Creation of a new laboratory rat in which both compounds and PMP22 can be tested at a level closest to human testing.

The creation of this new laboratory model will provide scientists with Schwann cells to test the already available drug compounds as discussed in Project 1, but the animals will also be used to actually test the candidate medicines as they are identified.

Continued on next page

Project 3

Characterization of the human PMP22 regions

This project will examine, in detail, the regulation of PMP22 in humans. What is known thus far comes from work with laboratory models. Logically, the next step is to study how the human PMP gene is “turned on” and causes an over-expression of that specific protein in CMT1A.

Following the completion of these projects, the CMTA will aggressively work to pursue any potential treatments arising from these trials, as well as

foster new strategies for treating other forms of CMT including 1X, and Types 2 and 4.

Initial studies focusing on CMT1A will inevitably be “translational,” directly impacting how we unravel the complexities of and ultimately treatments for other forms of CMT.

A vision of the CMTA is to promote collaborative work on an international level for developing effective therapies for CMT. To this end, an additional goal of the CMTA would be to develop partnerships with organizations such as the NIH and the MDA who share this vision.

SUPPORT THE STAR INITIATIVE



As the newly elected Chairman and President of the CMTA Board of Directors, I am extremely fortunate to be assuming the leadership of an organization that has such a solid foundation.

As a person with CMT, I am equally pleased to announce that the CMTA is now going full speed ahead with a new research initiative called the “Strategy to Accelerate Research (STAR).”

The goals of the STAR initiative are ambitious: We expect to develop therapies for the three most prevalent types of CMT within the next five years and to be able to reverse symptoms in some patients within ten years.

And these goals are attainable, though it will take an extraordinary commitment from the scientists, the Board of Directors, and everyone who supports the work of the CMTA.

To fund the STAR, we will need to raise \$10 million over the next five years.

With that in mind, our next fundraising appeal will be directed entirely to the STAR initiative. When you receive our request, please give generously. If you prefer to donate now, you can do so on the website, or by sending a check or credit card gift to the office. Additionally, you can remember the CMTA in your will or donate a gift of stock.

Together, we can change the face of CMT—forever!

A handwritten signature in blue ink, appearing to read 'Patrick Livney'. The signature is fluid and cursive, with a large initial 'P'.

Patrick Livney
Chairman and President of the CMTA Board of Directors

GET INVOLVED...



ASK THE DOCTOR

Dear Doctor,

I was recently prescribed Citalopram hydrobromide 20 mg. This is being prescribed for my depression following my wife's death. I am afraid to take this because of the possible effect it might have on my CMT. Can you advise me?

The Doctor replies:

Citalopram (Celexa) is an antidepressant in a group of drugs called selective serotonin reuptake inhibitors (SSRIs). There is no known link to neuropathy or worsening of CMT, but the drug does have a number of side-effects, as do all antidepressants. The drug can be quite helpful in grieving patients, but the prescribing doctor should address this patient's specific concerns.

Dear Doctor,

My husband has CMT and atrial fibrillation. Last year they put him on Rythmol. It was quite apparent that it affected his CMT. Then, they switched him to flecainide. In the last 6 months, his condition has really gone down. Tremors are getting bad, spasms in his legs, and his speech is getting difficult when he is excited. Is there any feedback on flecainide?

The Doctor replies:

His change on Rythmol (propafenone) is very intriguing. There are only 2 known and reported cases, both in 1995, of neuropathy resulting from this drug. There have been no subsequent reports, and the side-effect is not listed in the drug company materials. But, there is no

experience at all with this drug and CMT patients.

Flecainide is not known to cause or worsen neuropathy or damage nerves, but the drug can affect muscle and nerve function. It is occasionally used to suppress muscle cramps and similar neuromuscular spasms, but it is approved to treat abnormal heart rhythms. Tremor is a recognized and reasonably common side effect of flecainide, and it could certainly worsen tremor in someone with tremor from CMT.

Dear Doctor,

Can a patient with CMT take red yeast rice instead of a statin drug, or would red yeast rice be considered the same possible risk as the statin?

The Doctor replies:

I have heard about patients taking red yeast rice to avoid medications. I know of no literature linking this biologic agent and neuropathy or myopathy. The primary concern is whether it works sufficiently against the cholesterol problem.

Dear Doctor,

I have just been diagnosed with CMT. I also have dermatitis herpetiformis. When I have an outbreak, I need to take dapsone. What are the possible effects on my CMT?

The Doctor replies:

Dapsone is known to cause neuropathy that can affect the motor (strength) fibers and is a risk and concern. However, the

risk must be weighed against the benefit of treating your skin disorder. Most patients take dapsone without incident. There are no significant data about dapsone in CMT patients, but both tend to affect the same type of nerve fibers. The dapsone dose and length of treatment are important factors. You should pay close attention to your strength if the treatment is used.

Dear Doctor,

My daughter sent me an article about alpha-lipoic acid as a nerve damage repairer. The article suggested 300 mg, two to four times a day. The article referred mainly to diabetic neuropathy, but I wonder if it would work for CMT neuropathy.

The Doctor replies:

Alpha-lipoic acid (thioctic acid) is an antioxidant that has been studied mostly in diabetes. There is some objective evidence that the agent reduces painful symptoms, but it is not clear if it prevents nerve damage. The agent has not been tried on CMT to my knowledge. The mechanisms of nerve injury differ between diabetes and CMT.

Dear Doctor,

Are cortisone shots in any way toxic to CMT patients?

The Doctor replies:

Quick answer: single shots are not a problem, but longer-term chronic steroids can affect the muscles, but not worsen the neuropathy. ✱



SUPPORT GROUP NEWS

Colorado – Westminster

On April 26, 2008, Elizabeth Ouellette, a CMTA board member and the support group liaison, will speak to the group. Her topic will be her personal quest and experience in pain management. The meeting will be held at Capabilities, a retail store, in Westminster.

Illinois – Chicago Area

On a February afternoon, about 60 CMT patients, family and supporters gathered to hear a presentation by Dr. Michael Shy, a preeminent neurologist and CMT researcher from Detroit, Michigan. Dr. Shy lectured on genetics, medication interactions, the Wayne State CMT clinic, and current CMT research including the high-dose ascorbic acid trials. Also in attendance was Patrick Livney, the president and chairman of

the CMTA. He spoke enthusiastically on the future of the CMTA with a particular emphasis on the new advanced research initiative, named STAR. The next support group meeting will take place on May 17 at 2 p.m. The location is yet to be determined. For those who missed the last

meeting and would like to receive notes from Dr. Shy's presentation, please contact Alan Pappalardo, alan@charcotmarie-tooth.org.

Pennsylvania – Johnstown

February's meeting was great! We had nasty weather and no speaker but a great turnout. Attendance at CMT support meetings is a real conundrum. We schedule great speakers, have beautiful weather and the turnout is poor. When there is no speaker and snow, an enthusiastic crowd shows up. Go figure. CMTers must enjoy a challenge or CMT has addled our brains!

Anne Bootman, our new support group co-leader, brought a DVD of the CMTA's new awareness effort. The DVD has short clips of local support group members for TV stations. The clips have appeared on various health and news shows. Thanks, CMTA.

News Flash: Conemaugh Health Systems through The John P. Murtha Neuroscience and Pain Institute will be hosting a CMT Family Patient Conference on May 29th, in Johnstown, and Dr. Shy will be the featured speaker. Call 814-269-5232 for more info. Maybe it will snow and boost attendance!

Pennsylvania – Philadelphia Area

On February 16, 2008, Barry Kirsch, from Canine Partners for Life demonstrated the services that a dog can be trained to

perform. Sage, the demonstration dog, showed the attendees how she can open doors, turn off lights, pull shirts, jackets and pants off, and retrieve dropped items. The dogs are certified so that they can accompany people on airplanes, into restaurants, and anywhere else a person might need to go. Two of the group members are already in the process of applying to receive a service dog.

After the presentation, Pat and Dana discussed the patient/family conference being scheduled for April 5, 2008, at the University of Pennsylvania, the new STAR research initiative, and 25th anniversary plans for the CMTA.

Washington – Seattle

Our guest was Elizabeth Ouellette, member of the Charcot-Marie-Tooth Association Board of Directors <http://www.charcotmarie-tooth.org/cmta/bod.php> and Support Group Liaison for the CMTA. Elizabeth brought Kimberly Snadow along, a young woman with CMT who hopes to work for the association in California in the near future. Elizabeth gave an interactive presentation on how to describe CMT, using simple props, to children, family, and friends. Moreover, we were delighted to hear about the CMTA's new research plans (STAR) and the awareness campaigns soon to hit the national arena. We all left motivated by the STAR research program and all the CMTA is currently accomplishing. ✨

Service dog Sage accepts a monetary contribution from Sierra Syktich.



CMT Support Groups

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

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: ladyblue123@att.net

California—San Francisco Bay Area/Santa Clara County

Place: Menlo Park Library
Meeting: Quarterly
Contact: Elizabeth Ouellette,
650-248-3409 (C)
650-559-0123 (H)
Email: elizabetho@pacbell.net

Colorado—Westminster

Place: Capabilities, Westminster, CO
Meeting: 10 AM – noon, Last Saturday of
every other month
Contact: Diane Covington
303-635-0229
Email: dmcovington@msn.com

Florida—Tampa Bay Area

Place: St. Anthony's Hospital,
St. Petersburg, FL
Meeting: Second Saturday of Feb,
May, Nov, with other special
meetings throughout the year
Contact: Lori Rath, 727-784-7455
Vicki Pollyea
Email: rathhouse1@verizon.net
v_pollyea@mindpspring.com

Georgia—Atlanta Area

Place: Cliff Valley School Library
2426 Clairmont Rd, NE
Meeting: Bi-monthly, Third Saturday
Contact: Sue Ruediger, 678-595-2817
Email: cmtasg_atlanta@
charcot-marie-tooth.org

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,
Beaumont 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: Bill Miller, 763-560-6654
Email: wmillier758@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

Nevada—Las Vegas

Place: West Charleston Library,
6301 West Charleston Blvd.
Meeting: Email for dates 1-3 PM
Contact: Mary Fatzinger
Email: cmt_suppgrupp_lvnv@yahoo.com

New York—Greater New York

Place: NYU Medical Center/
Rusk Institute, 400 E. 34th St.
Meeting: Second Saturday,
12:30-2:30 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 Children's Hospital
Meeting: Bimonthly, Jan, March, May,
Sept, and Nov; 3rd Saturday
Contacts: Beverly Wurzel, 201-224-5795
Eileen Spell, 732-245-0771
Email: craneomatic@verizon.net
espell@optonline.net

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

Place: Raleigh, NC
Meeting: Quarterly
Contact: Susan Salzberg,
919-967-3118 (afternoons)
Betsy Kimrey
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@roadrunner.com

Pennsylvania—Philadelphia Area

Place: CMTA Office,
2700 Chestnut St., Chester, PA
Meeting: Bi-monthly
Contact: Pat Dreibelbis or
Dana Schwertfeger,
800-606-2682
Email: info@charcot-marie-tooth.org

Virginia—Harrisonburg

Place: Sunnyside Retirement
Community, Sunnyside Room
Meeting: Bi-monthly, Second Sat. 1-3
Contact: Anne 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

Washington—Tacoma

Place: Moore Library, 215 S.56th St.
Meeting: Bi-monthly
Contact: Carol Hadle
Email: heandme@q.com

SPOTLIGHT ON SUPPORT GROUP LEADERS

Lori Rath, Florida

Why did I start a support group? First of all, there wasn't one here in Florida. I had experience as a volunteer, working five years with the mentally challenged and physically handicapped. I always have to be involved in something: Church, Community, or Social Groups. I am a people person.

My childhood was full of action I enjoyed Girl Scouts, dance, acrobatics, ice skating, roller skating, and bicycling.

My first job was as a secretary in a large factory in Buffalo, New York. I was a lousy typist! My ambition was to get out from behind the desk and travel. I was hired by an airline company and worked at the ticket counter, made reservations and was also a flight attendant. A flight attendant has to do a lot of standing and walking. I also visited many, many cities, walking all the way. However, at this time in my life, I did not have any problems with my legs.

I even met my husband in an airport. He worked for a different airline. So, together, we did the traveling that I always longed to do. This is how I was able to see much of the world, and we plan to keep on taking trips in the future.

We have five children and at this time none show signs of CMT. At the age of 52, I became aware of the first signs

of CMT in me. It started with weakness in my hands, and then I found that I was tripping over nothing. One therapist told me that my physical problems were all in my mind (she said that she trips too).

As a support group leader, I get great satisfaction in supply-



Dr. Michael Shy visited Lori Rath and Vicki Pollyea's Tampa Support Group.

ing information to people with CMT through the speakers we have been fortunate to have. At the first meeting in November of 2004, Dr. Michael Franklin, my neurologist from the MDA clinic, spoke. We've had many other speakers: Vicki Pollyea, OTR, (my co-leader) spoke on "How to do More and Hurt Less." Other presenters we have had are a representative from Athena Diagnostics, a hand therapist, an orthotist, physical therapists, a speaker on "Emotional Perspectives on Coping," a nutritionist and a rehabilitation seating specialist.

I've had surgeries on my feet, including having bunions removed. My hammertoes have been straightened on my right

foot, and my right ankle has been reconstructed. There are five pins in it to keep it straight. Right now, my left foot is beginning to get curled toes. And my left ankle is in bad shape, but I will not go through the rehabilitation of six months in a cast and wheelchair, followed by walking casts.

My orthotist is preparing a new AFO to support my left ankle. The best shoe for me to accommodate an AFO is New Balance athletic shoes. They have roomy space for the toes and good gripping soles. My hands are weak and my handwriting is awful. A hand therapist gave me a brace, but that just caused blisters, as it was made of plastic and was designed to help me hold a pen and write better. I have so many problems picking up small objects, buttoning buttons, holding a sewing needle, etc. On a positive note, I can still tie my shoes!

Yes, I feel guilty that I may have passed this genetic disorder on to my children, but so far none show signs of CMT. I have three brothers, two who are very affected, and much older than I. I have a cane, but I only have to use it occasionally.

My motto is that I am not a handicapped person, but a person with a handicap. I feel that the support of my husband and family keeps me going. CMT has affected my legs and my hands, but not my spirit! ❄

SHAUN DAILEY*(continued from page 6)*

I will go on and inspire others with CMT to become active instead of sitting and letting this disorder get the best of them. I feel like I am a pioneer for athletes with CMT. My mother and sister, who also have this disorder, are growing closer everyday to becoming more active. I am training and pushing the envelope everyday to make it to the next level. My long-term goal is to accomplish an Ironman Triathlon, which is

a 2.4-mile swim, a 112-mile bike ride and a full marathon—26.2 at the end. I run 4 to 5 days a week, and my dream of accomplishing my goal is looking better everyday!

I have a desire to help make people aware of CMT. Racing and being at the events is the only way to really get the knowledge out about CMT. Since CMT is genetic, I worry that my two boys may have this disorder also, so my efforts are not only for myself but to prove to my boys that if they have CMT that they can do *anything*

in the world they want to with no limits. I want them to say, "If daddy can do it, then so can I!" This message is for you too, whether you are a parent of a child with CMT or a person who personally has this disorder; there are no limits and I have proven this! I am here to help, to encourage, and to inspire! If you need my suggestions, encouragement, or advice, please feel free to contact me, I want you to succeed, too!!

—Shaun Dailey, 770-856-3504
woods racer@bellsouth.net
shaun.dailey@cowetaschools.net

JAM-C Is a Potential Correction for Myelin Problems

A sticky molecule previously linked to inflammation also helps seal vital insulation around peripheral nerves, making it a potential target for new drugs against nerve disorders, scientists said on Thursday.

The latest research suggests the molecule, known as JAM-C, could be a key player in regulating the way nerves work.

In genetically modified mice without the adhesion molecule, the myelin insulation sheath protecting nerves deteriorates and the animals experience faulty nerve firing, muscle weakness and a shortened stride, researchers reported in the journal *Science*.

The team also found that nerves of patients with certain peripheral nerve disorders had defective JAM-C.

Taken together, the findings suggest the molecule is a key player in regulating the structure

and function of peripheral nerves and its malfunction may cause a number of illnesses.

JAM-C, which was discovered only recently, is already being studied as a target for new medicines involved in inflammation and as a possible route to fight cancer, since it seems to help tumors form new blood vessels.

"This finding opens up yet another area that this molecule should be investigated in — but it's very early days," Sussan Nourshargh, professor of microvascular pharmacology at Barts and The London School of Medicine, said in an interview.

Nourshargh made the discovery of the molecule's role in peripheral nerves by accident, while investigating blood vessels. Her team then collaborated with scientists at Imperial College London, University College London, Cancer Research UK

and the University of Geneva to advance the work further.

There are more than 100 kinds of peripheral nerve disorders affecting approximately one in 20 people. They often afflict people with existing diseases like diabetes and lupus, a chronic autoimmune disease.

Symptoms include numbness, pain, tingling, muscle weakness and sensitivity to touch. Problems often start gradually and steadily get worse.

Nourshargh said the new molecule was not found in the central nervous system and was therefore unlikely to play a role in multiple sclerosis.

JAM-C seems to work by sealing off the insulation in the critical gaps between so-called Schwann cells, which produce the myelin layers that wrap around nerve cells. *

Reporting by Ben Hirschler, reprinted from Reuters news service.

WRITE TO US!

Pat Dreibelbis,
Editor,
pat@charcot-marie-tooth.org

Dana Schwertfeger,
Director of Member
Services
dana@charcot-marie-tooth.org

The CMTA Report is published by the Charcot-Marie-Tooth Association, a registered non-profit 501(C)(3) health organization. © 2008, 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.

IN MEMORIUM

One of the Early Geniuses in the Study of Peripheral Nerves

Professor P.K. Thomas died on January 25th at the age of 81. He was a member of the CMTA's Medical Advisory Board, wrote a chapter in the *Physician's Guide to CMT* and was highly regarded by the CMT community as a founding father of the study of the disorder.

Born and brought up in Wales, P.K., as he was almost universally known, was proud of his Welsh roots. He was the eldest of three boys born to a family of timber importers based near Swansea in South Wales. He was a very bright child who, to the disgust of his more normal siblings, found time to teach himself to play the piano when he should have been doing school work, and still come out top in his exams. When he graduated, he moved to London to study Medicine at University College London. Luckily for him, the head of the department of anatomy at UCL was the noted anatomist JZ Young. P.K. was awarded a first-class honours degree in anatomy in 1947.

He chose to study the peripheral nervous system rather than the brain and quickly made the subject his own beginning in 1949 with a paper on internodal

length in fish which made an important contribution to understanding how nerves lengthen as an organism develops. This was followed by an MBBS in 1950 and a prestigious medal in clinical surgery. In the early years while training in medicine, he was very involved in exploring the correlation between nerve structure and function as measured by nerve conduction studies.



P.K. Thomas
1927-2008

He learned electron microscopy with David Robertson, a famous electron microscopist in Montreal from 1961 to 1962, and he set up an EM laboratory in Maida Vale Hospital in London on his return to a clinical post. His clinical interest in peripheral neuropathy was thus supported by electrophysiologic and pathologic studies at both the light and ultrastructural level. The attention to detail that made him such a good scientist and editor, however, never hindered his ability to see the larger picture.

A longstanding interest in hereditary diseases received a great boost with the development of modern molecular genetics in the 1980s, especially as he was by then married to Anita Harding who matched

him in intellect and the ability to work amazingly hard. They were a fantastic team and made valuable contributions to the understanding of genetic neuropathies. His ability to overcome the hurdles life can produce was tested when she died young of cancer in 1995. He then became very involved in studying the genetic neuropathies of the Roma (gypsies). He was fortunate to get Sam Ponsford as an M.D. student in 1998 and married her in 1999. She cared for him devotedly after his stroke in 2001 and gave him some fun in his last years.

Somehow he managed to fill every waking moment with first-class research and clinical activities and still have time to enjoy music, wine, travelling, boar hunting, etc. The departmental Christmas parties during his years as head were famous!

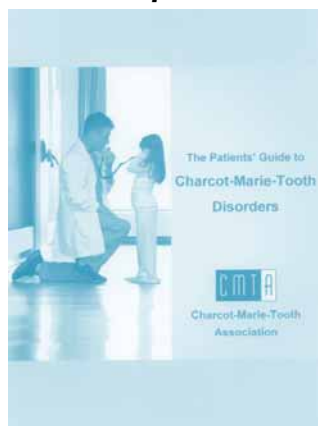
He was married 3 times, first to Mary in 1952 (who died in 1977) with whom he had 2 sons, Adrian and Nicholas (and 6 grandchildren), then to Anita, and lastly to Sam, who survives him. Even in old age and infirmity, he still had a twinkle in his eye and was adored by all the nurses who cared for him.

P.K. Thomas was unique and there will never be another like him. He will be sadly missed.

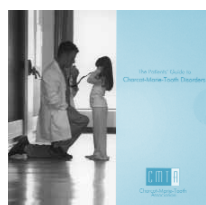
—Dr. Rosalind King
Dept. of Clinical Neurosciences
Institute of Neurology
University College London

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

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.



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
Lefluonamide (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 US)
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

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