1983-2008

WE CELEBRATE 25 YEARS: LIVING OUR VISION OF A WORLD WITHOUT CMT

THE CMTA Report

SEPTEMBER/OCTOBER
2008

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

Charcot-Marie-Tooth Association

OUR MISSION:

To generate the resources to find a cure, to create awareness, and to improve the quality of life for those affected by Charcot-Marie-Tooth.

OUR VISION:

A world without CMT.

EMAIL CMTA AT:
info@charcot-marie-tooth.org

ISSN #1067-0181 ✻ Vol. 23, No. 5

CMT Centers of Excellence to Open in January, 2009

BY PAT DREIBELBIS

The CMT North American Database was established in 2001 to house historical, clinical, and neurophysiological data on patients with Charcot-Marie-Tooth disorders. To date, over 800 patients have submitted their information to the database, and scientists and physicians have used the information housed there to identify medications that exacerbate CMT, to recruit patients for the ascorbic acid clinical trials, and to study genotype-phenotype correlations. Unfortunately, the current database is limited in the quality of its clinical data because a large number of physicians have evaluated the patients, and there are inconsistent neurological examinations and neurophysiology data.

To address these problems and improve the usefulness of the database, the database will become the North American CMT Network, with all patients being evaluated at one of six national centers: Wayne State University, the University of Pennsylvania, the University of Washington, Johns Hopkins University, the University of Texas Southwestern, and the University of Rochester.

Goal one in the establishment of these Centers is to ensure that patients are evaluated in a uniform and high-quality manner at one of the six Centers, each located in a distinct region of the US and led by a Principal Investigator (PI) with clinical expertise in peripheral neuropathy and experience with patients who have CMT. Patients currently enrolled in the Database will be given the option to be evaluated at the Center closest to their home. The PIs will offer their current CMT patients the opportunity to be enrolled in the Network. Quantitative scoring of the patient's impairment will be done by the use of the CMT Neuropathy Score (CMTNS) and the Neuropathy Impairment Score (NIS), and these results will be added to their clinical evaluations. The Network will then have a thoroughly evaluated pool of patients to use for future natural history studies and clinical trials.

> The Centers will provide high-quality, uniform evaluations for patients across the U.S.

(continued on page 2)

More friends join the CMTA's Circle, *see page 12*

INSIDE:

Herb Beron joined the CMTA Board in July.

Transitions

The CMTA is pleased to announce the addition of Herb Beron to its Board of Directors. Herb is excited at the direction the CMTA is taking and is anxious to be a part of all that it hopes to accomplish through STAR, the strategy to accelerate research. As he said, "When our daughter Julia was diagnosed with CMT 2E, my wife and I decided that we needed to become actively involved with the CMTA. Two years ago, we formed "TeamJulia" to participate with Steve O'Donnell in The Swim for the Cure, and since then we have raised in excess of $150,000. I received a BBA in Finance from Emory University in 1986, and I am currently a Senior Vice President for Citigroup Smith Barney. It is my heartfelt goal that the CMTA achieve its stated mission: "to find a cure, to create awareness, and to improve the quality of life for those affected by Charcot-Marie Tooth."

***

The CMTA also announces the resignation of its Executive Director, Charles Hagins. Charles worked for the CMTA for seven years and was largely responsible for the grants from the State of Pennsylvania that helped to launch the awareness campaign throughout the state and featured Coach Joe Paterno, Penn State Head Coach and College Football Hall of Fame Inductee. Under his tenure, the CMTA launched STAR, the strategy to accelerate research, a program that is hoped will bring treatments, therapies, and eventually cures for some forms of CMT. ✻

CENTERS OF EXCELLENCE

(continued from page 1)

A second goal of the new Centers will be to obtain DNA from the patients evaluated there and to bank the DNA at Indiana University, which currently houses the database. DNA from patients with known mutations will be available for haplotype analysis or to investigate modifier genes that could be responsible for differences in severity among patients with the same genotype (e.g., the same mutation). For the remaining patients, DNA samples will be available for gene identification.

A third goal will be to establish a scoring system for quantifying impairment in young children with CMT. The CMTNS and the NIS work well with patients ten years and older, but the scoring systems do not work in young children and infants. In this new program, a number of scoring systems used to evaluate children with neuromuscular diseases will be combined into a Neuromuscular Team CMT Evaluation (CMTE). The CMTE will contain data (nerve conduction, quantitative motor, and sensory scores) that are also components of the CMTNS, so this will facilitate transition into the CMTNS as patients age.

The Centers of Excellence will be funded by both the Charcot-Marie-Tooth Association and the Muscular Dystrophy Association. These Centers have been chosen because of the quality and experience of the doctors who will serve as the Principal Investigators. Dr. Michael Shy sees 1200 patients of all ages at the Wayne State CMT Clinic; Dr. Thomas Bird follows 400 CMT families with more than 1000 affected members at the University of Washington; Dr. Steven Scherer has more than 150 CMT patients in his care at the University of Pennsylvania, and Dr. Richard Finkel sees 100 children with CMT at the Children's Hospital of Philadelphia; Dr. Ahmet Hoke has 100 CMT patients at Johns Hopkins; Dr. David Herrmann has 50 patients with CMT at the University of Rochester, and Dr. Susan Iannacone sees 150 children with CMT at the University of Texas Southwestern. Taken together, over 2400 patients at the six sites are currently available as potential members of the CMT Network.

The establishment of these Centers bodes well for making the diagnosis of CMT and its future treatment uniform across the country. ✻

SAVE THE DATE

West Coast CMT Patient/Family Conference—November 8, 2008

BY ELIZABETH OUELLETTE

I am delighted to announce that on November 8, 2008, the CMTA will hold a Patient/Family conference in Palo Alto, California. The last CMT West Coast conference was approximately 10 years ago, so this much-needed event, designed to educate both medical professionals and patients alike, is an opportunity not to be missed. My hope is that each participant will invite personal physicians, friends, and family members to the conference where they will meet others with CMT and become more familiar with the disease. Attendees will learn more about STAR, our innovative research endeavor that is working to develop CMT treatments, and a possible cure, within 5 to 10 years.

This exciting event will feature experts in the field of Charcot-Marie-Tooth disorders, who will discuss a variety of topics crucial to understanding this progressive neuromuscular disease. Moreover, every attendee will have the chance to ask questions pertaining to CMT, on a one-to-one basis with many of the specialists before, during, and/or after the conference.

I am excited to announce that our speakers will include Dr. Michael Shy, Director of Detroit's Wayne State CMT Clinic and his team of dedicated CMT specialists. These specialists include Sean McKale, well-respected orthotist, Carly Siskind and Shawna Feely, a team of dynamic genetic counselors, and Dr. Rosemary Shy, CMT clinic pediatrician.

In addition to the above, several of the CMTA's Board members will be present, including Patrick Livney, Chairman and President of the CMTA's Board of Directors. Both Pat Dreibelbis, Director of Patient Services, and Dana Schwertfeger, Director of Member Services, who work at the CMTA's national headquarters in Pennsylvania, will be present to answer questions, receive commentary, and bring attendees up-to-date on current CMTA-related activities. Furthermore, I have asked representatives of the MDA, CMTUS, and Shriners Hospitals to speak about the many services offered to a CMT patient.

Attendees can ask questions pertaining to CMT of the specialists, one on one.

Other CMT specialists will be asked to give presentations as well, so, as November approaches, we will keep you posted as to additional developments. The price for the day is $15 for CMTA members and $20 for non-members, which includes a light continental breakfast, lunch, and afternoon snacks.

The conference will be held at the Mitchell Park Community Center in Palo Alto, CA, and is scheduled to begin at 8:30 am and end at 5:30 pm, with an hour break for lunch. Everyone will receive a token gift from the CMTA for completing a conference survey, and those who bring a friend, physician, or family member who is not already enrolled in the CMTA's database, will receive a free CMT t-shirt.

We are working very hard to make this conference beneficial and successful by raising community awareness, educating the CMT patient, and bringing hope to all those attend. People in West Coast states will receive invitations in the mail, with a conference registration form. You can also register at: www.cmtausa.org/paloalto. If you have specific questions, please call the CMTA: 1-800-606-CMTA ext.103, or email conference@charcot-marie-tooth.org. We look forward to receiving your registration and meeting you in November at this memorable event! ✻

The Kinds of CMT…Lessons in Cell Biology

BY STEVEN SCHERER, MD, PHD, UNIVERSITY OF PENNSYLVANIA

This is the first in a series of essays, collectively intended to explore key concepts regarding the structure and function of peripheral nerves, and to relate these ideas to the different kinds of CMT. My goal is to inform interested readers in these matters, so that they can further explore these topics for themselves. I encourage readers to use the following websites, all of which are excellent and informative: Online Mendelian Inheritance in Man (OMIM; www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=OMIM), the Mutation Database of Inherited Peripheral Neuropathies (www.molgen.ua.ac.be/CMTMutations/), the Neuromuscular Disease Center of Washington University (http://www.neuro.wustl.edu/neuromuscular/), and GeneTests (http://www.genetests.org). There is an outstanding and comprehensive book, **Peripheral Neuropathy**, *and an excellent group of reviews published in the journal* NeuroMolecular Medicine *that can be downloaded as PDFs from the CMTA website, but these sources are written for neurologists, and are thus too detailed for most readers. I recommend the book published by the CMTA,* **The Patients' Guide to Charcot-Marie-Tooth Disorders**, *as a good starting point. The current version of the popular on-line encyclopedia, Wikepedia, is not consistently reliable, or even comprehensive enough to be informative on these topics. I have tried to write at the level of a college biology class, highlighting key concepts. I apologize in advance for my mistakes and any confusion I generate. Unless it proves to be too demanding, I hope that I will be able to answer questions regarding these essays by email (sscherer@mail.med.upenn.edu). I welcome your suggestions.*

THE KINDS OF CMT

CMT is the eponym for Charcot-Marie-Tooth disease. The name itself reflects that these physicians were the first to describe individuals with inherited neuropathy (~1885). Hereditory sensory and motor neuropathy (HMSN) is a widely used alternative name, and peroneal muscular atrophy is an older term that is fading away. The term CMT is properly used to describe a neuropathy that affects sensory and motor axons. Patients with the disease are otherwise well. The designation CMT should not be used for inherited diseases that strongly affect other organs, such as the brain; these are syndromes in which neuropathy is typically an unimportant element (there are at least 100 such syndromes). The terms congenital hypomyelinating neuropathy (CHN) and Dejerine-Sottas neuropathy (DSN) are used to describe severe neuropathy with a clinically recognized onset in infancy or before three years of age, respectively. As the name indicates, sensory (and variably autonomic) neurons/axons are affected in hereditary sensory and autonomic neuropathy (HSAN), with relative or complete sparing of motor neurons/axons. Conversely, in hereditary motor neuropathies

Fig. 1:

The "central dogma" of molecular biology. *Genes are comprised of DNA (which is double-stranded). DNA is transcribed into RNA (which is single-stranded), which in turn is translated into protein. DNA can self-replicate.*

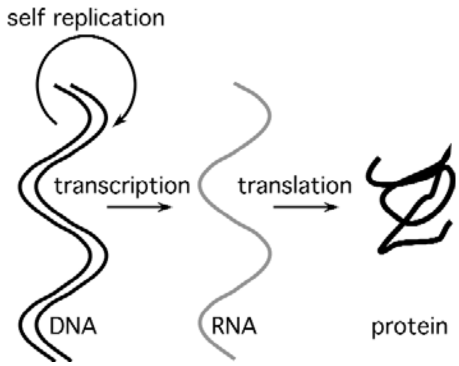
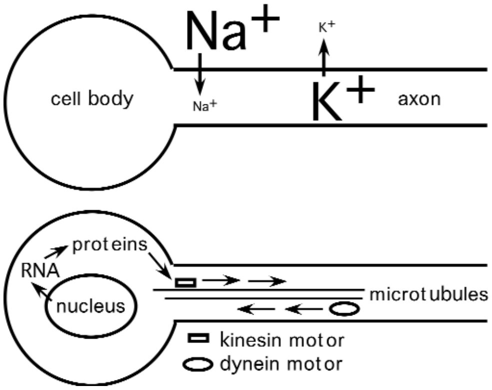


Fig. 2:

The anatomy of neurons and axons. *The upper panel is a schematic of a neuron, which is composed of its cell body and axon. Electric signals (action potentials) are generated by the rapid influx of sodium ions (Na+) into the axon, and travel down the axon. The lower panel shows that proteins are synthesized in the cell body and shipped down the axon attached to kinesin motors.*



(HMN), motor but not sensory axons are affected.

All kinds of CMT share in common the finding that nerves are progressively damaged over time. There are many kinds of CMT (about 50 identified so far, and no end in sight; Table 1), and each kind can be conceptualized as a molecular lesion of a single gene. Because genes encode proteins (*Fig. 1)*, each mutated gene generates a malfunctioning protein. Exactly how these proteins malfunction is not easily determined: this would require that one already knows the function of its normal counterpart, and this is typically not the case. One key generalization was made by Albert Aguayo, Garth Bray, and their colleagues in 1977: inherited demyelinating neuropathies are caused by the effects of mutant protein in myelinating Schwann cells. Conversely, it appears that inherited axonal neuropathies are caused by the effects of mutant proteins in neurons, especially their axons. The clinical correlation of this dichotomy is that CMT can be separated into two kinds—the "demyelinating" forms (CMT1 and CMT4) and the "axonal" forms (CMT2, autosomal recessive CMT2, HSAN, and HMN).

NEURONS, AXONS, AND AXONAL TRANSPORT

To understand the effects of mutations, we need to understand axons and their myelin sheaths. Most neurons have a single axon, which is physically contiguous with the neuronal cell body *(Fig. 2)*. Axons are the part of a neuron that is specialized for conducting electric signals. The axonal membrane contains channels for the two main ions that created bioelectricity—sodium ions (Na+) and potassium ions (K+). Both Na+ and K+ flow down their gradients when the appropriate channels are open. Because the concentration of Na+ is higher outside the axon (in the blood) than inside the axon, Na+ flows into the axon when Na+ channels are open. This instantaneous flow of Na+ into the axon generates the bioelectric signal called the action potential. Depending on the size of axon, and whether

(continued on page 6)

Table 1:

The molecular genetics of nonsyndromic inherited neuropathies. Dominant disorders are bolded; OMIM numbers are listed in parentheses; Asterisks denote genes for which genetic testing is available (http://www.genetests.org).

DISEASE	LINKAGE OR GENE
CMT1 (autosomal or X-linked dominant demyelinating)	
HNPP (162500)	*PMP22** (601097)
CMT1A (118220)	*PMP22** (601097)
CMT1B (118200)	*MPZ** (159440)
CMT1C (601098)	*LITAF** (603795)
CMT1D (607687)	*EGR2** (129010)
CMT1X (302800)	*GJB1** (304040)
DI-CMT: Dominant Intermediate CMT	
DI-CMTA (606483)	10q24.1-25.1
DI-CMTB (696482)	*DNM2* (602378)
DI-CMTC (608323)	*YARS* (603623)
CMTX3 (302802)	Xq26
CMT2 (autosomal dominant axonal/neuronal)	
CMT2A1 (118210)	*KIF1B** (605995)
CMT2A2 (609260)	*MFN2** (608507)
CMT2B (600882)	*RAB7** (602298)
CMT2C (606071)	12q23-24
CMT2D (601472)	*GARS** (600287)
CMT2E (162280)	*NEFL** (162280)
CMT2F (606595)	*HSPB1** (602195)
CMT2G (608591)	12q12-q13.3
CMT2 (604484)	3q13.1
CMT2-P0 (118200)	*MPZ** (159440)
CMT2L (608673)	*HSPB8** (608014)
CMT4 (autosomal recessive demyelinating neuropathy)	
CMT4A (214400)	*GDAP1** (606598)
CMT4B-1 (601382)	*MTMR2** (603557)
CMT4B-2 (604563)	*MTMR13* (607697)
CMT4C (601596)	*SH3TC2** (608260)
CMT4D (601455)	*NDRG1* (605262)
CMT4F (145900)	*PRX** (605725)
HMSN-R (605285)	10q23.2
CMT4H (609311)	*FGD4* (611104)
CMT4 (605253)	*EGR2** (129010)
CMT4J (611228)	*FIG4* (609390)
AR-CMT2: Autosomal Recessive Axonal Neuropathy (CMT2B)	
AR-CMT2A (605588)	*LMNA** (150330)
AR-CMT2B (605589)	19q13.1-13.3
"CMT2K" (607831)	*GDAP1** (606598)
Congenital AR axonal neuropathy	5q deletion
HSAN: Hereditary Sensory and Autonomic Neuropathy	
HSAN1 (162400)	*SPTLC1** (605712)
HSAN1B (608088)	3p22-24
HSAN2 (201300)	*WNK1** (605232)
HSAN3 (223900)	*IKBKAP** (603722)
HSAN4 (256800)	*NTRK1** (191315)
HSAN5 (162030)	*NGFB** (162030)
Primary erythromelalgia (133020)	*SCN9A** (603415)
HMN: Hereditary Motor Neuropathy	
HMN I (606595)	
HMN II (158590)	
HMN II (608634)	*HSPB8** (608014) *HSPB1** (602195)
HMN III (607088)	11q13
HMN IV	
HMN V (600794)	*GARS** (600287) *BSCL2** (505158)
HMN VI/SMARD1 (604320)	*IGHMBP2** (600502)
HMN VIIA (158580)	2q14
HMN VIIB (158580)	*DCTN1** (601143)
HMN/ALS4 (602433)	*SETX** (608465)
Congenital distal SMA (600175)	12q23-q24

Abbreviations: HNPP (hereditary neuropathy with liability to pressure palsies; SMARD1 (spinal muscular atrophy with respiratory distress type 1)

THE KINDS OF CMT

(continued from page 5)

the axon is myelinated, the action potential travels at 1 meter/second (in small, unmyelinated axons) to 80 meters/second (in large, myelinated axons). Conversely, because K+ is higher inside the axon than outside the axon, K+ tends to flow from the axon when K+ channels are open. An energy-requiring "pump" (Na+/K+-ATPase) maintains the low intracellular levels of Na+ and the high intracellular level of K+.

Axons are in continuity with the neuron cell body. This continuity is essential: if an axon is physically separated from the neuron cell body, then it will degenerate distal to the site of separation. Part of the reason that axons depend on their neuron cell bodies is that most of the biomolecules in axons are made in the nerve cell body, then shipped down the axons in a process known as axonal transport. Axonal transport requires a motor (made of kinesin) that can transport molecules and a road (made of microtubules). Mutations in the gene that encodes one of these motors, kinesin 1, causes a very rare form of CMT2 (CMT2A). There is a different motor, dynein, that transports cargo in the opposite direction—to the nerve cell body. Mutations in a component of this dynein motor, dynactin-1, cause a hereditary motor neuropathy (HMN VIIB).

Because axons are such a long way from their cell body, their energy needs must be provided locally. Mitochondria generate most of the energy used by most cells, including neurons and their axons. Thus it stands to reason that mutations in two genes that encode various components of mitochondria cause inherited neuropathies—*MFN2* (CMT2A2) and *GDAP1* (CMT4A). In addition, mutations in a large number of other genes encoding other components of mitochondria cause inherited neuropathies that are part of more complex syndromes.

Axons have an internal skeleton composed of microtubules and even more importantly, neurofilaments. Neurofilaments are composed of three subunits, termed heavy, medium, and light. Dominant mutations in *NEFL*, the gene encoding the light subunit, cause an axonal neuropathy (CMT2E). Mutations in the genes that encode two different heat shock proteins may cause dominantly inherited axonal neuropathy (CMT2F, CMT2L, and HMN II) by disrupting the metabolism of neurofilaments. Finally, it should be noted that recessive mutations in the gene that encodes gigaxonin also cause a neuropathy. Gigaxonin interacts with microtubule-associated proteins, and the absence of gigaxonin function results in grossly enlarged axons. ✻

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 ☐ American Express
Card # ____________________________
Exp. Date ____________________________
Signature ____________________________

Gift Given By:
Name: ____________________________
Address: ____________________________
____

An Amazing Night in Rhode Island

BY PAT DREIBELBIS

On Friday, July 25, 2008, at 9:30 am, Dana Schwertfeger and I loaded up my car with video equipment, newsletters, brochures, and pens and headed out for Rhode Island. The trusty MapQuest directions assured me that the drive would be a manageable five hours and we would arrive in West Warwick, RI, long before the scheduled dinner/dance for Grace's Courage Crusade. What MapQuest always forgets are things like road construction, massive amounts of plain-every-Friday traffic around New York City, and accidents. Stopping only for lunch and two bathroom breaks, we finally made it to Rhode Island around 5:25 pm. If you do the math, you will figure out that our simple little five-hour trip took almost 8 hours, and it was anything but pain-free.

We arrived so late that there was no time for video set-up and checks or other last-minute preparations. We had to change into dinner clothes and get to the event. I would be less than honest if I said that our spirits were high as we left the hotel. But, I would be equally remiss if I failed to remark that our spirits were lifted almost immediately upon entering the West Valley Inn and seeing Marybeth Caldarone, her daughter Grace, her husband Chris, and the organizer of the event, Tonia Hassell. There was so much love and enthusiasm that it was impossible not to be infected by it.

An amazing crowd of over 200 people had purchased tickets for the dinner and dance. The evening began with a table of appetizers and drinks and the opportunity for everyone to look over the huge table of silent auction items and the equally huge table of desserts that would be part of a "dessert war" after the meal, where tables would bid to have the baked good of their dreams delivered to their table for consumption. If I'm ever asked where all the bakers of yesteryear have gone, I will quickly respond that they all live in Rhode Island. The cakes, the brownies, the pies, and even the cookies were home made and fabulous both in appearance and taste (I can speak personally regarding a brownie and a wonderful cookie that found their way to my table.)

There was a buffet dinner and speeches by Patrick Livney, Chairman of the Board of the CMTA, Dr. Louis Weimer, Columbia Presbyterian, Dana Schwertfeger, who played the STAR DVD for the crowd, a representative from the Ronald McDonald House in Rhode Island, and myself. Tonia's husband, Lew Hassell, presented a slide show of pictures of Chris, Marybeth, and Grace to music that left very few dry eyes. Then, Chris and Grace had a father-daughter dance to the music from Steven Curtis Chapman called Cinderella, and whatever dry eyes there had been were dry no longer.

Between the dessert was the silent auction, the live auction, and the balloon sales, along with the dinner tickets, the night grossed almost $30,000. The success of this fundraiser can be attributed to several things: the immense love and respect the community has for Marybeth and Grace Caldarone; the intense work ethic of Tonia Hassell and her committee of supercharged men and women; and the higher purpose of the whole event, namely finding therapies, treatments, and eventually a cure for CMT. ✻

Chris Caldarone danced with his "princess" Grace on her special night.

PATIENT PROFILE

Allen Pearson: Nature Photographer, Artist

I was born with CMT but not diagnosed until age 20. CMT for me means nerve pain in my legs, arms, and hands; loss of sensation in my hands and feet; loss of muscle in my legs, feet, hands, and arms; deformed feet with hammer toes; and the inability to fully use my hands; and plenty of difficulty walking, turning my ankles easily. For the past eight years, I've worn braces to improve my stability and energy. On occasion, I've had to use a wheelchair when I'm on my feet for long periods. It sounds depressing, but it really isn't. Sure, I have had my moments or days just like anyone else would. However, a few years after my diagnosis, I decided that I had to make the best of what I have, and I needed to focus on the beauty and positive aspects of each day. I determined I wasn't going to let it get me down.

> "I photograph aspects of nature which are often missed by most people."

Soon after my diagnosis, God spoke to my heart saying that He would be my strength, not my physical body, if I would trust Him. I read the Scripture, "But those who hope in the Lord, will run and not grow weary, they will walk, and not be faint" (Isaiah 40:31, NIV). This scripture has been an important part of my life, and God has kept His promise to me.

Concerned about side effects of medications, I decided to pursue the natural or holistic approach to CMT. I have found a primary care physician, a sports medicine doctor, an orthopedic specialist, a certified massage therapist, a chiropractor, a deep water aerobics instructor, and physical therapists, all who are continually instrumental in helping me to feel the best I've ever felt.

When I have issues with CMT, I focus on things in my life that bring me happiness: my faith, my wife, my dog, my cat, family, classical music, nature, and photography. Focusing on these has helped me through the roughest times, and they remind me that I am blessed and loved.

My passion for nature led me to become a gardener and to create several annual and perennial gardens and a backyard wildlife habitat (which is creating a garden to feed wildlife). To get my backyard wildlife habitat certified by the National Wildlife Federation, I had to include pictures of my work. As I photographed, I became more interested in the aspects of nature, and my interest in photography began to take hold.

My photography skills began getting noticed, and I received many compliments. So in 2006, I started a small photography business. I photograph aspects of nature which are often missed by most people and I work to portray them as fine art. I offer two photography services, Garden and Dog/Cat. I sell my work as fine art in the form of prints and art note cards. You can view my work at www.allenpearsonsphotos.com. If you are in the area of one of my shows, I would enjoy meeting you. ✲

GIFTS WERE MADE TO THE CMTA

IN MEMORY OF

Ethelene Breuer
Mr. and Mrs. John A. Stuller

Jim Cahill
Mr. Erhard Gersbach

James Cargill
Mr. and Mrs. William T. Ankney
Mr. and Mrs. R. Chessman
Mr. Dennis W. Culley
Ms. Gwendolyn D. Delonnay
Mr. and Mrs. Edward F. Dollar
Mr. and Mrs. Robert Facko
Mr. and Mrs. Al Gruenewald
Mr. and Mrs. Laurence E. Houghtaling
Mr. and Mrs. Douglas M. Jewell
Mr. and Mrs. Don Johnson
Mr. and Mrs. Thomas C. Kizis
Mr. and Mrs. James H. Kurt
Mr. and Mrs. William J. Langley
Mr. and Mrs. J. Libke
Ms. Mary W. McFarland
Mr. and Mrs. Billy Moorefield
Mr. Marvin Spear and Mr. William Hart
Mr. and Mrs. Larry Nolin
Mrs. Patricia A. Purtee
Mr. and Mrs. William Rice
Mr. and Mrs. Kenneth W. Robinson
Ms. Christine Seebon
Mr. John W. Wozena

John W. Carr
Mrs. Cynthia J. Church

Betty Chow
Mr. Henry Chow

Susan Elmer
Mr. and Mrs. Philip De Spirito

Amy H. Hedstrom
Mr. and Mrs. Thomas C. Ainsworth
Mr. and Mrs. Francis W. Blane
Ms. Linda D. Brand
Mr. and Mrs. Dorrence B. Brucker
Mr. and Mrs. Michael Carver
Mr. and Mrs. Jay Cave
Mr. and Mrs. Neil E. Conklen
Mr. and Mrs. Kenneth Cunningham
Ms. Wilma Doggett
Mr. and Mrs. John R. Gehlbach
Mr. and Mrs. Thomas L. Jackson
Mr. and Mrs. David G. McAfee
Mr. and Mrs. James Miller
Mr. and Mrs. Evan A. Minder
Mr. David Tomlin and Ms. Sheryl Samuelson
Mr. and Mrs. David W. Nelson
Mr. and Mrs. Donald J. Ogden
Mr. and Mrs. James F. Schlouch
Ms. Erma Schussele
Springfield Area Arts Council
Mr. and Mrs. Robert D. Thomas
Mr. and Mrs. Robert L. Umbach
Ms. Lauralee Zimmerman

Stephen E. Jumper
Ms. Cynthia J. Donley
Ms. Helen J. Ford
Ms. Elizabeth J. Livingston
Mr. and Mrs. Bob Marioni
Mr. and Mrs. Mickey Mitchell

Patrick W. Kerr
Mr. and Mrs. John M. Deal
Mr. and Mrs. Fred W. Hedge, Sr.

Robert Kitchens
Ms. Juanese Calkins

Howard A. Parker
Ms. Sharon L. Snook

Teresa Pegazzani
Mrs. Anna Maria Park

Daniel P. Peoples, Jr.
Mr. Jerry Haney
Ms. Joyce Shirey
Ms. Verna Thompson
Ms. Joann Wisor

Anita Ray
Mr. Jess D. Ray

James Smith
Mr. Christopher Palermo

Sarah Wien Robin
Ms. Leanore R. Cohen

IN HONOR OF

Anna den Boef
Mr. and Mrs. Philip J. Brown
Holy Family Catholic School

Jerry Donovan
Ms. Elaine Donovan

Renee Gelman
Mr. Leon Gelman

Marlene Goodman
Mrs. Stacey Bernson

Debbie Hagen
Mr. Michael Bohrer

Mr. and Mrs. Emil Klimah
Mr. and Mrs. Richard Betten
Ms. Heidi D. Conradt-Fish
Mr. and Mrs. David L. Hartwig
Mr. James Hartwig
Mr. and Mrs. Arthur E. Hartwig
Mr. and Mrs. Ernest Holtzman
Mr. Stan Wilcinski and Ms. Elizabeth Wilezynski
Mr. Michael Pappalardo, Sr.
Ms. Mary Ann Schneider
Mollie and Linda Siska
Ms. Mary G. Slawinski
Mr. and Mrs. Robert O. Superek
Mr. and Mrs. John Thurau
Mr. Richard Thurau
Mr. and Mrs. Stephen D. Voss

Charlene Landry
Ms. Kami K. Landry

Margaret Lee
Mrs. Elizabeth Brown

Julie Leonard
Mrs. Shirley C. Weiner

“Captain” Pat Livney
Mr. Carter Auslander
Mr. Thomas Brown
Mr. Michael T. Clune
Mr. Chris Doerr
Mr. Mickey Hamano
Mr. Clay Hamano
Mr. John Henderson
Mr. Brian Hill
Mr. Paul Hoffman
Mr. John Koffel
Mr. Frank Laskaris
Mr. D.J. McCullough
Mr. Mark Miller
Mr. Jess Ray
Mr. Bradford Smart
Mr. Hugh Williams
Mr. Jim Young

Tyler Ray Lopez
Mrs. Jean A. Moore

Evan Jacques Isaac Marouf
Ms. Jody Kyle
Windmill Point Park

Alan Pappalardo
Mr. Michael Imrisek

Douglas and Kathleen Sanford
Mrs. Maria Barone
Mr. John Cabral, Jr.
Mr. and Mrs. John Chakuroff
Mr. and Mrs. Richard Cleary
Mr. and Mrs. Mario E. Dessi
Ms. Kathryn J. Halliday
Ms. Rachel J. Hatch
Ms. Virginia M. Hatch
Ms. Nancy R. Lancaster
Mr. and Mrs. Harold J. Messenger
Mr. and Mrs. Dave Mixer
Mrs. Kim Mowbray
Mr. and Mrs. Fred Paris
Mr. Gary R. Sanford
Mr. and Mrs. Eric and Denise Stockton/McMahon
Mr. and Mrs. Andrew Williams

Lauren Sanford
Mr. and Mrs. Fred Paris

Mr. and Mrs. Matt Valentine
Ms. Rita A. Zadurski

LaRue M. Wadford
Mrs. Cheryl A. Stevens

Sadie Zanzuri
Ms. Jane Vayman

CMT IN THE NEWS

Newsmakers Spread Awareness of CMT

THE INTERNET SERVICE, CONTACTMUSIC.COM announced that Julie Newmar, a legend for playing Catwoman in the 1960's Batman series, was diagnosed six months ago with CMT. Her long legs were once insured for $1 million, but she now holds on to people to avoid falling down. Newmar, 74, says, "Today I walk, but very slowly. My balance is also affected. People might think I've had too much to drink, but I never drank, smoked, or took drugs my entire life. When I'm out in public, I grab on to some charming fellow who can steady me."

THE CLEARFIELD COUNTY, PA PAPER, THE PROGRESS HOME, reported on Nancy Syktich and her daughter, Cierra Kephart, who has been diagnosed with CMT. Cierra's daily routine includes a 15-step exercise regimen, and she attends therapy sessions three times a week. Every six months she travels to Philadelphia for checkups and fittings for new braces. Cierra began walking late at 14 months and fell a lot. Doctors in their home area couldn't diagnose the problem and simply said that Cierra "walked funny." Despite the disorder, Cierra is a normal little girl who faces all the challenges of an 11-year old. Instead of riding the bus to school, she is picked up by a van and an aide helps her throughout the day. But she has great friends who understand her problem and look out for her safety. They all like to go swimming, play on a trampoline, play videogames, and have sleepovers.

THE NEW YORK TIMES published an article entitled, "Has Trumpet, Will Surprise," about John McNeil, one of the best improvisers working in jazz, who performs in a small restaurant in Brooklyn, called Biscuit. Mr. McNeil has dealt with Charcot-Marie-Tooth disease from childhood, wearing braces at times from his legs to his neck. There were days when CMT kept his fingers from doing what he wanted them to and he even has been affected in his diaphragm and tongue. By 1997, he could not extend the fingers in his right hand, which he uses for fingering his instrument. He was recently fitted for some handsome new custom-made finger braces that allow him to curve his fingers better. *(Editor's note: They are sterling silver and come from the Silver Ring Splint Company, www.silverringsplint.com)* Mr. McNeil has largely hidden his disease from the public, though now he says it might be useful for people to know about it—so they can see that it's no big deal.

John McNeil uses his finger braces to allow him to play the trumpet.

HOUSTON COMMUNITY NEWSPAPERS ONLINE reported on Cathy Powers who has tremors or what she called "wilders." When she experienced a numbness in her foot, she knew right away what it was. She was diagnosed with CMT at the age of 37. The history of the disorder goes back in her family to the 1800s. "My family called it "dead foot" disease and "the Wilders" because my great-grandmother's last name was Wilder," said Cathy. She has an active job

(continued on page 16)

THIRD ANNUAL CMT GOLF TOURNAMENT A HUGE SUCCESS

Patrick Livney (right), shown here with this twin brother Roland, has organized golf tournaments for the past three years to raise money for CMT research. This year's event netted almost $140,000 for the research fund and the STAR initiative.

Being Part of the Greater "Whole"

BY ALAN PAPPALARDO

At the age of three, I was diagnosed with CMT. By the age of 23, the diagnosis was revised to include my subtype, 2A. As I grew older, my disease progressed to the point where it took away most of my walking and fine-motor skills. At the age of three, there is little one can do with the devastating news of a CMT diagnosis, but upon learning of my updated diagnosis, I chose to use my time and skills to help defeat CMT by joining the Charcot-Marie-Tooth Association staff. I credit many people and groups for giving me the strength to keep fighting against what has steadily taken so much away: my friends, my family, and equally importantly, the organizations dedicated to our cause: the Muscular Dystrophy Association (MDA) and the CMTA.

I have felt the influence of the MDA in my life for as far back as I can remember. They helped pay for my leg braces, clinic visits, and many other services I utilized. My parents greatly appreciated the help MDA gave me. As a family we fundraised and we disseminated information, but my fondest memory is being selected to be the Chicagoland regional poster child.

During second and third grade, I traveled Northern Illinois doing a wide variety of tasks: I spoke at golf outings, appeared in commercials, gave interviews, and even joined the cast of the Chicago MDA telethon. As a child, I was limited in the scope of my actions. I remember the excitement of meeting celebrities, the drain of constant promotion and travel, and even the occasional trinket from a contributor. But even then I knew I was there to put a face on our fight, to show the world what CMT is and that if we are going to beat it, we all have to pitch in.

Dr. Shy gave me something worth more than anything I could have imagined: *hope.*

MDA taught me by example what was needed to be done to eradicate CMT and helped give us patients a quality of life through programs designed to bring people with CMT together in safe, open environments. Through the eight years I attended the MDA summer camp program, I saw firsthand how prevalent diseases like CMT are, and I also saw that I was not alone.

The best gift that summer camp gave me was the opportunity to meet people like myself, dealing with a disease and trying hard to prosper. One man I met at camp, Patrick Livney, stands out because of the significance he had on my development as a person and in life. Patrick is also a CMT patient and now he's the Chairman of the CMTA Board of Directors. He was my counselor when I was eight, and over the course of a wild week filled with swimming (new to me) and violently boisterous wheelchair hockey, I made a friend and an inspiration. Meeting Patrick was the single most significant memory I have from all of my years of summer camp.

Flash forward to 2007, I had recently completed college at Washington University in St. Louis. I got a call from Patrick Livney telling me about the CMTA and its exciting research initiative. He had me visit the CMTA-sponsored clinic in Detroit's Wayne State University run by Dr. Michael Shy.

On a snowy Friday in March, I made my way to Detroit for a doctor's visit that would change my life. Never before had I been witness to a collection of physicians and people so informed and dedicated to CMT. After a battery of exams and lengthy questions, Dr. Shy was able to give me something worth more than anything I could imagine: hope. He identified the type of CMT I have and told me that with that knowledge, I could choose to have a family without the fear and guilt of passing on CMT.

I realized that I need to make everyone know that if they can help, then they have to do so. Whether it is becoming a member of the CMTA, donating money, writing articles or participating in support groups, we must each do anything and everything to bring an end to the legacy of CMT. ✻

The CMT "Circle of Friends"

The people who have become involved in the CMTA's Circle of Friends program are making an important contribution that will benefit all of us as we work to find a cure for CMT.

If you'd like to start a Circle of Friends, please call us 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. Here are some new circles as well as some new contributors to older circles.

Jules Ochoa

I would like to thank all my family and friends for donating money to my Bar Mitzvah project, for funding of the CMTA's research. We raised almost 1,300 dollars! And every one of those dollars counts! I would also like to say that even with CMT, I am playing football this season, which goes to show that if you want to do something badly enough, things will work out and you will find a way to do it.

—Jules Ochoa

Jules Ochoa's Bar Mitzvah Gift

Jules decided that he would ask his family and friends to send whatever they would have given him as a Bar Mitzvah gift to the CMTA's research fund.

Contributors to Jules' Bar Mitzvah:
Rabbi and Mrs. Kerry Baker
Mr. John W. Bederman
Ms. Anita Shapiro Braun
Mr. and Mrs. Nick Carter
Central TX Pediatric Ortho & Scoliosis Surgery
Mr. and Mrs. Daniel H. Cook
Mrs. Melanie F. Cox
Dennis Fagan Photographer
Mr. Robert M. Franklin
Ms. Sarah Gaertner
Mr. Rich Gramann
Ms. Diane S. Graves
Mr. Alan Greenberg
Mr. Caleb Gross
Mr. and Mrs. Dennis Gross
Mrs. Linda R. Halbreich
Mr. Marc Jacobs
Mr. and Mrs. Neil B. Kimmelman
Mr. and Mrs. Byron P. Kocen
Ms. Eileen Ladd
Ms. Elizabeth Maupin
Ms. Lynny D. Moore
Mr. and Mrs. David H. Ochoa
Mr. and Mrs. Michael E. Ochoa
Dr. Rona Pogrund
Mr. and Mrs. Myron Radwin
Mr. Reuven Robbins
Mr. and Mrs. Irving B. Rose
Cantor Robbi Sherwin
Mr. Ted Toth and Ms. Karen Siegel
Mr. and Mrs. Jerome F. Vitemb

Contributors to Reagan's Quest:
Ms. Tammy Derynda
Mrs. Beverly A. Foti
Ms. Sue Gouvion
Mr. Kari A. Haluska
Mr. and Mrs. Dave Madsen
Mrs. Carrie Perock
Mr. Randall Rathburn
Mr. and Mrs. Gene D. Rueter
Mr. and Mrs. Mark R. Stultz

Reagan Stultz's parents will run a haunted house fundraiser in October.

Additional Contributors to Amy's American Tears Project:
Mr. Peter deSilva
Mr. and Mrs. Dennis Dowd
Mr. and Mrs. George Gagnon
Mr. and Mrs. Donald R. Sorelle
Mr. Anthony Troy

Additional Contributors to Grace's Courage Crusade:
Ayco Charitable Foundation
Mr. and Mrs. James C. Dakin
Mr. and Mrs. John Oberle
Ms. Angela D. Scungio
Mrs. Nancy W. Sherman
Dr. David N. Silvers
United Way of RI

Additional Contributors to Team Julia '08:
Citi Global Impact Funding Trust, Inc.
Kingdon Capital Management
Kraft Foods, Inc.
Mr. and Mrs. David J. Sarney

The CMT "Circle of Friends"

Kaitlin Dineen's "John-a-Thon"

Kaitlin Dineen surprised her father John by organizing a walkathon in his honor. See *Letters* below, for his reaction.

Contributors to John-a-Thon:
Mr. and Mrs. J. Kim Abdallah
Mr. and Mrs. Gary A. Allcorn
Mr. and Mrs. Harvey I. Black
Mr. and Mrs. Dennis E. Boitnott
Mr. and Mrs. William L. Brister
Mr. and Mrs. David S. Brown
Mr. and Mrs. David S. Brown, Jr.
Ms. Meghan N. Brown
Ms. Olivia S. Cameron
Mr. and Mrs. Hector A. Colindres
Mr. John Dombrowski
Megan and Tom Duffy
Mr. and Mrs. James A. Ermon, Jr.
Mr. and Mrs. David A. Gardoni
Mr. and Mrs. David M. Geary
Ms. Meryl S. Hankins
Mr. and Mrs. Richard E. Hart
Hauerland's
Mr. and Mrs. M. W. Hays, Jr.
Mr. and Mrs. Brian D. Herman
Mr. and Mrs. David J. Herman
JDC/Firethorne. Ltd
Mr. and Mrs. William P. Martin
Mr. and Mrs. Kevin McCarthy
Ms. Shannon J. McConnell
Mr. and Mrs. Michael Miarka
Mr. and Mrs. Larry Nix
Mr. and Mrs. Geoffrey Ondrus
Mr. and Mrs. James W. Pierce
Ms. Rhonda Reyes
Mr. and Mrs. Donald R. Robinson
Mr. and Mrs. Sam R. Scheiner
Seahorse Deepwater Technology, Inc.
Mr. and Mrs. Gene Sparkman
Mr. and Mrs. Matthew D. Steger
Ms. Sandra Troxell
Ms. Yolanda M. Velasquez
Ms. Jennifer L. Webb
Mr. and Mrs. Michael L. Williams

Kaitlin and John Dineen

LETTERS

Dear CMTA,

My daughter, Tanneal was diagnosed with CMT Type 1 in 2005. Since then, she has undergone surgery to lengthen her Achilles tendons. This has allowed her to put her feet flat on the ground, but many normal childhood activities still elude her. She did undergo physical therapy following her surgery with little progress. In late 2006, after trying several sports in our community, Tanneal showed an interest in swimming. She has since flourished as a member of the Boonslick Heartland YMCA Blue Marlins Swim Team. Although she is not the best swimmer on the team, she can keep up with the other kids, which is something that she has never been able to do in the past. Swimming has proven to be a wonderful tool to help her because the rigorous workout in the pool is low impact. She swims 2 hours a day, 3 days a week, 40 weeks per year.

Not very long ago, I founded a grass roots organization to request that our City Council consider the construction of an indoor recreational and competition aquatic facility, so that our community can impact more children with similar needs and improve our community infrastructure as a whole. I am currently in the process of exploring grant opportunities.

I want to reiterate the benefits of swimming to patients with CMT. I would love to see more research done in relation to this particular sport and its benefits to those with CMT.

—T.H. by email

Dear CMTA,

I am writing to thank you for the support given my daughter, Kaitlin, in organizing the Walk-a-Thon we staged on May 24th here in Katy, TX. She completely stunned me in arranging the project as a complete surprise. I've been blessed throughout my life in spite of dealing with CMT. Kaitlin knew that an event such as this would be a gift I will cherish the rest of my life. We only hope that our donations will help find a cure for the generations to come.

—John Diveen, TX

WRITE TO US!
Pat Dreibelbis,
Editor,
pat@charcot-marie-tooth.org

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

SUPPORT GROUP NEWS


Dennis Devlin and Yohan Bouchard met at the Philadelphia area support group and shared mutual concerns over CMT-related back issues.

California – San Francisco

The group held its annual picnic at the home of leader, Elizabeth Ouellette on September 6, 2008. The major serious topic of conversation was the upcoming patient/family conference on November 8, 2008. See article about it on page 3.

Colorado – Westminster

The last meeting was held on August 30, 2008 and featured Dr. Ronald Kramer, neurologist and recognized authority on sleep apnea. The next meeting of the group will be held on October 25, 2008, and will feature Sean McKale, an orthotist from Wayne State University. The meeting time is from 10-noon.

Massachusetts – Boston

The “kick-off” meeting of the group was September 27, 2008, in the Shapiro building at the Beth Israel Deaconess Medical Center. Mark Boxshus, new leader of the group, envisions the support group as a “home” for patients, friends, and family members. At this first meeting, the group was able to ask CMT medical questions of Dr. Andrew Tarulli, a neurologist from the Beth Israel Deaconess Medical Center. The group was also treated to a song from Amy de Silva and a viewing of the STAR video which features her.

Michigan – Ann Arbor

The inaugural meeting of the group was a great success, with 35 in attendance. The meeting involved getting to know everyone and setting up goals for the coming year. On September 13th, the group heard Dr. Michael Shy discuss the characteristics of CMT and the ambitious Strategy to Accelerate Research. On October 18th, the guest speakers will be Carly Siskind and Shawna Feely, genetic counselors, and Sean McKale, an orthotist, all from Wayne State.

Nevada – Las Vegas

The Las Vegas support group will meet on Saturday, November 22nd from 1-3 PM The guest presenter will be Daniel Antonino, who is the owner of Performance Physical Therapy. He will be discussing various at-home activities that can be done to keep our bodies healthy. Call Mary Fatzinger at 702-369-6095.

New York – Horseheads

The group met on August 13, 2008, and had two new members in attendance. Because there were new members, the questions lasted for two hours, but the group felt they eased the new members’ minds regarding the progress of CMT.

The next meeting will be October 8, at 7 pm at the Horseheads Free Library, South Main Street, Horseheads, NY. The speaker will be Dr. Nilay Shah, a neurologist.

Pennsylvania – Johnstown

The group held its regular bi-monthly meeting Saturday, August 23, 2008, at the John P. Murtha Neuroscience and Pain Institute. The guest speaker was Dr. Sharon Plank, who discussed healthy and healing eating and living. Dr. Plank prepared and distributed several delicious, healthy treats, including chocolate-covered fresh blueberries which were “to die for.”

The next meeting is scheduled for Saturday, October 25th, from 1-3 pm at the Neuroscience and Pain Institute.

Pennsylvania – Northwest

The group met on September 13, 2008, at the Blasco Memorial Library to discuss CMT items from the Internet, articles from professional journals, and scientific articles. Because of a lack of CMT specialists in the area, the group usually holds meetings without invited speakers and chooses to share success stories and information they have gathered along the way in living with CMT.

Pennsylvania – Philadelphia Area

On Saturday, August 16, 2008, the group enjoyed a presentation by Support Group Liaison Elizabeth Ouellette on pain management techniques. The group also saw the STAR video featuring Amy de Silva and received the new neurotoxic drug cards. Betsy Chandler also spoke about the upcoming “Jazzed for a Cure” evening of jazz entertainment to be held in Villanova, PA, in March 2009. The group will meet on October 11, 2008, to hear an orthopedic surgeon discuss options for correcting foot and ankle deformities. ✻

CMT Support Groups

Support Group Liaison: Elizabeth Ouellette, 1-800-606-2682, ext 107

Alabama—Birmingham
Contact: Dr. Dice Lineberry, Calls only 205-870-4755
Email: dkllrl@yahoo.com

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: San Mateo 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
Contact: Lori Rath, 727-784-7455
Vicki Pollyea
Email: rathhouse1@verizon.net
v_pollyea@mindspring.com

Georgia—Atlanta Area
Place: Cliff Valley School Library
2426 Clairmont Rd, NE
Meeting: Third Saturday of every other month
Contact: Sue Ruediger, 678-595-2817
Email: susruediger@comcast.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, Beaumont Branch
Meeting: Quarterly
Contact: Martha Hall, 502-695-3338
Email: marteye@mis.net

Massachusetts – Boston Area
Place: Beth Israel Deaconess Medical Center
Meeting: Bi-monthly
Contact: Mark Boxshus, 781-925-4254
Email: MarkB_CMTANE@mac.com

Michigan – Ann Arbor
Place: Great Lakes Regional Training Center
Meeting: Monthly
Contact: Tammy Mayher, 517-451-8471
Email: dmayherjr@netzero.com

Minnesota—Benson
Contact: Rosemary Mills, calls only 320-567-2156
Email: rrmills@fedtel.net

Minnesota—Twin Cities
Place: Call for location
Meeting: Quarterly
Contact: Bill Miller, 763-560-6654
Email: wmiller758@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: carole.haislip@sbcglobal.net

Nevada—Las Vegas
Place: West Charleston Library,
6301 West Charleston Blvd.
Meeting: Email for dates 1-3 PM
Contact: Mary Fatzinger
Email: cmt_suppgroup_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)
Email: nabosmom@gmail.com

Ohio—Greenville
Place: Brethren Retirement Community
Meeting: 4th Thurs. of April, July and October
Contact: Dot Cain, 937-548-3963
Email: Greenville-Ohio-CMT@woh.rr.com

Oregon—Portland Area
Place: 1008 NE Division, Suite B
Gresham, OR
Meeting: Quarterly
Contact: Debbie Hagen
Email: hagen84@yahoo.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
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

COMING SOON: Baltimore, Maryland

ID of Protein that Prevents Myelin Formation Could Have Implications for CMT Treatment

FROM *QUEST*, SEPT.–OCT. 2008; VOL. 15 (NO. 5)

Scientists in the United Kingdom and Italy have found that a protein called c-jun keeps cells associated with nerve fibers from maturing and producing myelin, a fatty sheath that insulates fibers and speeds transmission of signals to and from nerve cells.

Kristjan Jessen at University College London and colleagues, who reported their findings May 19 in the *Journal of Cell Biology*, say it's likely the normal role of c-jun is to push myelin-making cells, known as Schwann cells, back to a more primitive state after nerves are injured. Schwann cells normally return to this earlier stage of development after injury, they note, as part of the process of nerve-fiber repair and regeneration.

However, in Charcot-Marie-Tooth disease (CMT), abnormal loss of myelination occurs, slowing nerve signals and leading to disability. Several forms of CMT, including the relatively common CMT1A and CMT1B, as well as other diseases, are characterized by abnormalities in myelination of nerve fibers.

The investigators say it will be important to determine whether c-jun is involved in causing these abnormalities. If so, they say, targeting c-jun might open new avenues for treatment. ✻

MORE NEWSMAKERS

(continued from page 10)

teaching toddlers and has to make adjustments, such as not standing too much and wearing herself out. As she said, "A long shopping trip is hard. I can't shop for two hours on my feet."

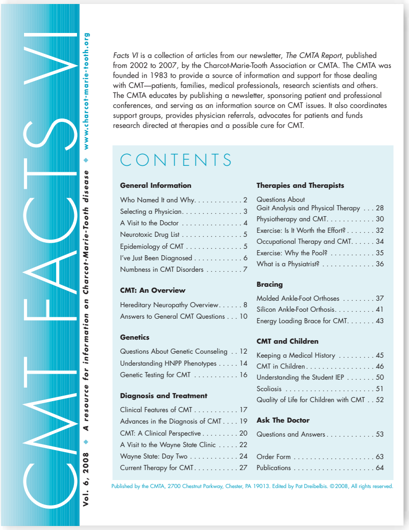
A COMMEMORATION OF THE LIFE OF P.K. THOMAS, former member of the CMTA's Medical Advisory Board and noted British neurologist, will be held on November, 25, 2008, at 5 PM at the Royal College of Physicians, Regents Park, London, United Kingdom.

THE MUSCULAR DYSTROPHY'S 2008 NATIONAL GOODWILL AMBASSADOR is Abbey Umali of Redlands, CA, an eight-year old with a form of Charcot-Marie-Tooth. Abbey is the first ambassador to hail from California in the program's 55-year history. She and her family will travel the country representing families afflicted with neuromuscular diseases served by the MDA. Abbey is the only child of Joel and Wendi Umali. Abbey has congenital hypomyelinating neuropathy, characterized by lack of coordination and balance, along with muscle weakness.

AN EXCERPT FROM BIO SMARTBRIEF announced that Claes Wahlestedt, a scientist with Scripps Florida, and Joe Collard, a business consultant, have established a new firm called cuRNA. The company will work on Scripps-licensed technology based on the potential of non-coding RNAs—small molecules that can increase or inhibit gene expression—to treat diseases and to serve as diagnostic markers.

THE PRESS ENTERPRISE reported on cyclist Anthony Zahn of Riverside, CA, who is competing in the Beijing 2008 Paralympic Games. The Paralympics are competitions for elite athletes in six disability groups. Zahn competes in a class of athletes who have locomotor disabilities, because he suffers from CMT. He will compete in four events. His favorite is the road race and he was quoted as saying, "I just want to see how far I can go and how many people I can beat, if anyone." Zahn, 33, is the owner of Anthony's Cyclery in Riverside's Canyon Crest Town Centre. ✻

CMT FACTS VI



CMT FACTS VI

Vol. 6, 2008 ◆ A resource for information on Charcot-Marie-Tooth disease ◆ www.charcot-marie-tooth.org

Facts VI is a collection of articles from our newsletter, *The CMTA Report*, published from 2002 to 2007, by the Charcot-Marie-Tooth Association or CMTA. The CMTA was founded in 1983 to provide a source of information and support for those dealing with CMT—patients, families, medical professionals, research scientists and others. The CMTA educates by publishing a newsletter, sponsoring patient and professional conferences, and serving as an information source on CMT issues. It also coordinates support groups, provides physician referrals, advocates for patients and funds research directed at therapies and a possible cure for CMT.

CONTENTS

General Information

CMT: An Overview

Genetics

Diagnosis and Treatment

Therapies and Therapists

Bracing

CMT and Children

Ask The Doctor

Published by the CMTA, 2700 Chestnut Parkway, Chester, PA 19013. Edited by Pat Dreibelbis.

is now available…

The sixth and newest edition of the CMT Facts Series has just been published and is available for purchase. It is 64 pages in length and is divided into sections on general information, genetics, diagnosis and treatment, therapies and therapists, bracing, CMT and children, and "Ask the Doctor."

The compilation of articles goes back as far as 2002 and captures the most significant articles from The CMTA Report from then to the present. Some of the more interesting articles involve numbness in CMT, HNPP phenotypes, current therapies for CMT by Dr. Michael Shy, exercise options, various types of bracing, IEPs for children, and the interesting and diverse questions posed to members of our Medical Advisory Board and answered by them in a ten-page section of the publication.

The cost for members is $12 and for non-members, $15.

(Please see next page for the entire CMT Facts series and other publications.)

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.

	QTY	COST	TOTAL
MEMBERSHIP (Free during 25th Anniversary year) Members have the option of receiving *The CMTA Report* in print, PDF via email, or both. Receive newsletter as: ☐ Print *or* ☐ PDF via email		FREE	
Receive both Print *and* PDF Newsletters		FREE	
The Patients' Guide to Charcot-Marie-Tooth Disorders [Print Format]		**members $10** nonmembers $15	
[CD Format]		**members $7.50** nonmembers $10	
CMT Facts I ☐ English ☐ Spanish		**members $3** nonmembers $5	
CMT Facts II ☐ English ☐ Spanish		**members $5** nonmembers $7	
CMT Facts III		**members $5** nonmembers $7	
CMT Facts IV		**members $8** nonmembers $10	
CMT Facts V		**members $12** nonmembers $15	
NEW! CMT Facts VI		**members $12** nonmembers $15	
A Guide About Genetics for CMT Patients *(No shipping and handling on this item only)*		**active members $4** nonmembers $5	
Teaching Kids about CMT…A Classroom Presentation *(1 hour DVD)*		**members $7.50** nonmembers $10	
CMT Informational Brochure, Neurotoxic Drug Card, and Letter to Medical Professional with Drug List		FREE	
Physician Referral List: States: ______ ______ _____		FREE	
Donation to the CMTA *(100% Tax-deductible)*			
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.

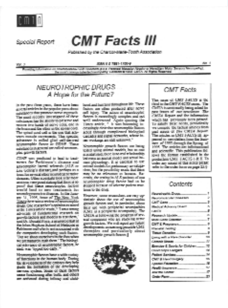
THE CMT FACTS SERIES: An Indispensable Collection of Articles about CMT

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

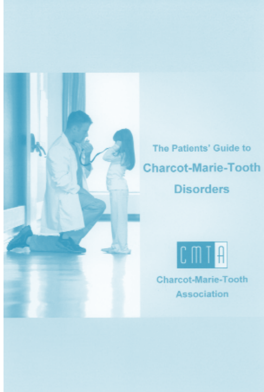
- Incurable, not untreatable
- CMT and pregnancy
- Prenatal testing
- Q & A about living with chronic illness
- Exercise & sports for children with CMT
- Patient services
- CMT hand surgery
- Special education
- Health insurance

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



The Patients' Guide to Charcot-Marie Tooth Disorders

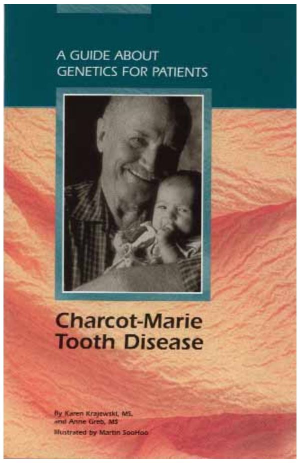
2008/178 pages—Now available in print and CD formats

The Patients' Guide is an excellent source of information and resources for patients, family members, and physicians. Topics include CMT in children, genetics and genetic counseling, orthopedic considerations, exercise, and current and prospective treatments for CMT.

Bonus features included on CD: *Charcot-Marie-Tooth Disease: An Overview* and *Charcot-Marie-Tooth Disease: A Guide for Physicians.*

PRINT FORMAT:
Nonmember Price: $15.00
Member Price: $10.00

CD FORMAT:
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

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

ASK THE DOCTOR

Dear Doctor,

I have been diagnosed with CMT for five years. The last two years, I have been free of pain and seem to have increased strength in my legs and hands. I have been taking Cymbalta. It seems to have worked for me. Has it worked for other patients? I'm not diabetic.

The Doctor replies:

The drug, Cymbalta, seems to work for all painful neuropathy, not just for diabetes, although that was its original purpose.

Dear Doctor,

I am a 58-year old male living in the Chicago area. I work from home and have a pretty sedentary lifestyle. Every winter I suffer the same problem. Even though my hands and feet are always cold, unless the ambient room temperature is 75 degrees or above, both my hands and feet perspire profusely. This phenomenon confuses me since the whole reason the human body perspires is to keep it from overheating through the process of evaporation of the perspiration. Therefore, I have two questions:

Is the excessive perspiration of my cold hands and feet a condition associated with CMT or is something else going on?

If it is associated with CMT, could you explain why it happens?

The Doctor replies:

I have conferred with two other CMT experts, and we are all puzzled. It is completely reasonable to think that CMT is causing the problem, depending on what type of CMT you have. In the rare disease called "cold induced sweating" (which is not really a form of CMT, but is a related variant), a low dose of a centrally acting antihypertensive medication, clonidine (0.1 mg twice a day) helped the sweating. You internist might try that. Alternatively, an anticholinergic medication may help.

Dear Doctor,

Today my internist asked me to begin taking either cerefolin or metanz. Are either of these helpful for CMT? Do they contain medications that would be harmful? Cerefolin contains L-methylfolate 5.6 mg, methylcobalamin 2 mg, N-acetylcysteine 600 mg. Metanx contains L-methylfolate 2.8 mg, pyridoxal 5-phosphate 25 mg, and methylcobalamin 2 mg. Thank you for your time.

The Doctor replies:

I looked up the two vitamin supplements. As stated, it appears that Metanx contains 25 mg of pyridoxal 5-phosphate which is a form of B_6. Excessive doses can cause neuropathy, but it is still controversial how much is too much. Most multivitamins contain 2 or 3 mg. Most toxicity cases are in the 100 mg/day range, but there are suspicions about toxicity with lower doses of 50 to 100 mg/day in some individuals. Twenty-five mg/day is probably fine, but it is much more than the recommended daily allowance.

Cerefolin appears to have no B_6, but includes other vitamins and one antioxidant that have no known toxicity on CMT. But it is unknown whether they have a benefit on neuropathy.

Dear Doctor,

My husband has CMT2 and has recently been diagnosed with hepatitis C. The doctor has determined it to be active, and we need to begin treatment. Is it safe for him to take Pegintron and ribivarin? Information will be greatly appreciated.

The Doctor replies:

The question is a bit complicated. Ribavarin is not associated with neuropathy, except at very high doses. Pegintron (peg-interferon) is rarely associated with neuropathy. However, hepatitis C infection is a much more common cause of neuropathy and a potentially serious disorder if left untreated.

Dear Doctor,

My 12-year old daughter has just been diagnosed with CMT1. She also has migraines and has been taking Pizotifen (Sandomigran) 0.5 mg, 2 tablets, for the past year. It may be pure coincidence, but her symptoms have certainly increased this past year and we are wondering if this medication is the cause?

The Doctor replies:

Pizotifen is not currently available in the US, but is available in some other countries. However, medications with similar effects (serotonin and antihistamine) are not known to cause or worsen neuropathy.

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

CMT PATIENT MEDICATION ALERT:

Definite high risk (including asymptomatic CMT):
Vinca alkaloids (Vincristine)

Moderate to significant risk:
Amiodarone (Cordarone)
Bortezomib (Velcade)
Cisplatin and Oxaliplatin
Colchicine (extended use)
Dapsone
Didanosine (ddI, Videx)
Dichloroacetate
Disulfiram (Antabuse)
Gold salts
Lefluonamide (Arava)
Metronidazole/Misonidazole (extended use)
Nitrofurantoin (Macrodantin, Furadantin, Macrobid)
Nitrous oxide (inhalation abuse or vitamin B12 deficiency)
Perhexiline (not used in US)
Pyridoxine (mega dose of Vitamin B6)
Stavudine (d4T, Zerit)
Suramin
Taxols (paclitaxel, docetaxel)
Thalidomide
Zalcitabine (ddC, Hivid)

Uncertain or minor risk:
5-Flouuracil
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*

CMTA
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