



**Charcot-Marie-Tooth  
Association**

**OUR MISSION:**

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

**OUR VISION:**

A world without CMT.

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**EMAIL CMTA AT:**  
[info@charcot-marie-tooth.org](mailto:info@charcot-marie-tooth.org)

## The CMTA Board of Directors Establishes \$350,000 Challenge Grant

BY ELIZABETH OUELLETTE

**A**re you up for a challenge? I hope you will jump on board and join us upon hearing this marvelous news: our Board of Directors is excited to announce the first-ever Challenge from the Board of Directors! For every dollar donated to the CMTA, our Board of Directors will match your gift on a one-to-one basis up to \$350,000!

At a recent meeting in Florida, the entire board brainstormed ways to convey its excitement and optimism regarding the new direction and promising future of the CMTA, and show its gratitude for all the fundraising efforts of past and current members, supporters, families, and friends.

Executive Director Charles Hagins explained that, "our intention is not only to express our appreciation for the generous donations large and small that the CMTA has received in years past, but also to demonstrate our unified board commitment to our new and ever-growing infrastructure, national expansion, and collaborative research plans."

Convinced of the untapped potential of the CMTA,



**Jason Steinbaum, Robert Kleinman, Patrick Torchia, Phyllis Sanders, and Steve O'Donnell all joined the Board challenge. Also contributing, but not pictured, are Gary Gasper, Patrick Livney, and Elizabeth Ouellette.**

board member Patrick Livney emphasized that "to achieve our long-and short-term goals, fundraising becomes imperative so that we can guarantee enhanced CMT awareness and increased research grants dedicated to finding treatments and a cure for CMT."

In order to raise part of the start-up costs associated with all of our new and innovative endeavors, Patrick proposed building upon the success of prior individual challenges by making a motion that every board member make a signifi-

cant pledge, which would then be pooled together in the form of a Board Challenge Campaign.  
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**BOARD CHALLENGE***(continued from page 1)*

paign. Patrick convincingly stated that “to demonstrate the board’s overall and total commitment, there is no better way than to put our money where our mouth is” and with these words, he received an overwhelming hands-down approval from each and every board member, making this challenge the largest on record in the history of the CMTA.

Moreover, Board Member Gary Gasper, whose 8-year-old son has CMT, is determined not to let research slide because of lack of funding. Gary reminds us that “the CMTA is at an exciting juncture, one where the upcoming collaborative and innovative research process will eventually create a non-stop source of funding for promising treatments without the long time delays with which we are presently faced.” Gary’s motivation stems in part from the belief that we will soon have real treatment options which will make the lives of anyone suffering from CMT better and more productive. As for those of us with children, Gary hopes that “we may be able to avoid and/or eliminate many of the problems and stigmas currently associated with growing up with CMT.”

Board Chairman Patrick Torchia, whose commitment to and financial backing of this organization has been exemplary over the years, is also fighting for our future generations. As Pat reflects upon the grand strides made over the past several years, he confidently affirms

that “the recent developments within our association will make it possible to successfully meet all of our aspirations and objectives.” Pat’s heartfelt desire, one which he is determined to fulfill, focuses on giving back to our kids and their families by providing hope for the future.

As for board member Steve O’Donnell, whose fundraiser “The Swim for the Cure” has been phenomenally successful, he believes that “the vision of finding a cure for CMT is much clearer today than it ever

assistance. In the past 5 years, the CMTA has grown into a respected agency that funds significant research projects and lobbies legislators to focus on CMT and promote our cause to fund significant research.”

Since I have been involved with the CMTA, I have had the opportunity to meet a lot of people who have CMT or have family members with this neuromuscular disease. In my role as support group liaison, I have been thoroughly impressed by the courage, stamina, and moti-

“The vision of finding a cure for CMT is much clearer today than it ever has been. More so than ever, we need to finance the exciting research and mission that the CMTA is undertaking.”

has been.” Steve appeals to each and every one of our supporters: “More so than ever, we need to finance the exciting research and mission the CMTA is undertaking.” Steve sincerely hopes that everyone will see our vision and help support us in meeting our goal.

Another fundraising board member, Robert Kleinman, has made a significant commitment to the challenge because of his experiences on the CMTA board and his interaction with the CMT medical community. As Bob said, “I am convinced that the CMTA is the best vehicle in existence today devoted solely to furthering CMT research and providing patient

vation of the individuals with whom I have come into contact. The more engaged I become, the more I want to assist the CMTA in all of its encouraging ventures.

My pledge toward this challenge demonstrates how strongly I feel about the ability of the CMTA and its researchers to discover new methods of treating CMT. As a member of such a resolute team, I feel enlivened by the contagious passion and enthusiasm among the members of this board. Moreover, this new spirit has spread, energizing our CMTA staff, our new development officer, Dan Merchant, and, of course, our

# MEMBERSHIP APPLICATION/ PUBLICATIONS ORDER FORM

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

	QTY	COST	TOTAL
<b>ANNUAL MEMBERSHIP DUES</b>			
Members have the option of receiving <i>The CMTA Report</i> in print, PDF via email, or both. Receive newsletter as: <input type="checkbox"/> Print <i>or</i> <input type="checkbox"/> PDF via email		\$40	
Receive both Print <i>and</i> PDF Newsletters		\$45	
Charcot-Marie-Tooth Disorders: A Handbook for Primary Care Physicians		<b>active members \$15</b> nonmembers \$20	
CMT Facts I <input type="checkbox"/> English <input type="checkbox"/> Spanish		<b>active members \$3</b> nonmembers \$5	
CMT Facts II <input type="checkbox"/> English <input type="checkbox"/> Spanish		<b>active members \$5</b> nonmembers \$7	
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CMT Facts V		<b>active members \$12</b> nonmembers \$15	
A Guide About Genetics for the CMT Patient (No shipping and handling on this item only)		<b>active members \$4</b> nonmembers \$5	
CMT Informational Brochure		FREE	
Physician Referral List: States: _____		FREE	
Letter to Medical Professional with Drug List		FREE	
<b>Contribution to CMT Research Fund</b> <b>(100% of contribution is used to fund research)</b>			
<b>Shipping &amp; Handling</b> (Orders under \$10, add \$1.50; orders \$10 and over, add \$4.50)			
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Check payable to the CMTA (US residents only; non-US residents, please use credit card or international money order.)

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**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.*

Executive Director, who recently commented, "I have never seen so much vitality, intensity and unity among board members, which is due in large part to all the innovative changes taking place within the organization."

With this being said, as members of the CMTA board, we are willing to lead but cannot carry the entire organization to its future goals alone. We need your help and commitment! Remember, if you take advantage of this matching challenge grant, your donation will be doubled, until we reach our \$350,000 target. Your future, as well as ours, is in your hands.

Long-time board member Phyllis Sanders describes this unique opportunity with two words: "Carpe Diem!" She stresses the importance of seizing the moment—which is now! Phyllis has observed the growing momentum created by our most recent research and awareness programs. This includes the statewide Pennsylvania awareness project, the engagement of our medical community to search for a cure and the interest the NIH has demonstrated by encouraging studies on CMT. In addition, Phyllis not only firmly believes that "we can do more," but is also persuaded that, "with additional funding, we will find a cure!"

Our hope is that you will join us in building a better, more efficient organization, one which will eventually provide the treatment options we have all been waiting for. There is no better time than the present to show your continued support, so when you receive the second quarterly appeal letter in the month of July, please give generously and help us meet this memorable challenge. \*



# CMTA Opens Satellite Office in Chicago

BY ALAN PAPPALARDO

The CMTA is expanding their operations to the Chicago area by opening a small office in the region where I will work. My name is Alan Pappalardo and I was diagnosed with CMT at the age of three. Recently, I traveled to Detroit's Wayne State University Clinic to establish a baseline on my condition, and to determine what form of the disease I had. With the help of the staff at the clinic, I now know I have CMT2A.

My life started on Chicago's south side, in November of 1983. What began as clumsiness and difficulties walking eventually led to a CMT diagnosis. From the first signs of physical abnormalities, my parents and sister worked tirelessly to teach me that I am no different from anyone else. When my condition worsened, and I chose to use a wheelchair, I vowed to live every day with a positive attitude.

From age eight through ten, I served as the Chicago Regional Poster Child for the Muscular Dystrophy Association (MDA). For seven years, I attended the MDA Summer Camps. My memories from those summers are filled with a happiness that comes from feeling completely free and unaffected by my disability. I raised tens of thousands of dollars and volunteered my time extensively for MDA.

However, my recent experiences with the CMTA have caused me to shift my support; I am now working to eradicate CMT.

In 1994, my family and I moved to the Chicago suburb of Naperville. During high school, my physical condition began to rapidly degrade, and I lost the



ability to walk long distances. I managed to graduate near the top of my class of 700 and won multiple honors. Daily, I fought off the pain and embarrassment of walking and endured the all-too-regular falls. However, I successfully walked every single day of my high school career.

One of my proudest moments was in 2006 when I earned my bachelor's degree from Washington University in St. Louis. While a student, I worked as a disability advocate for the university, leading training sessions, helping students, and lecturing to classes.

In addition to attending a university far away from my home, I made the difficult decision to convert to a wheelchair. My walking had become severely

limited and I realized that if I were to succeed in living independently, I had to change. I often describe CMT as "frustratingly debilitating." With the use of a wheelchair, a new world of mobility opened up to me.

We are living in a time of promise and excitement for those of us with CMT. Society and law have begun to embrace the changes necessary for the success of the disabled community. Innovative gene research has brought a new hope to CMT patients for a cure.

My work with the CMTA is one of the proudest challenges of my life. With my help, the CMTA is expanding into the Chicago region. A new office will serve as the focal point for this initiative; support groups, fundraising opportunities, and local CMT information will be just some of the benefits for CMT patients and their families.

Over the past year, I have observed, interacted, and directly benefited from the CMTA. What I found from this organization was a progressive agenda dedicated to helping eliminate CMT. Everyone involved is genuinely excited to fight CMT and tirelessly works with a passion that is unparalleled. I would like to invite everyone who reads this to feel free to contact me, get involved, and realize that our future is one of progress and hope. \*

# Researchers Discover Gene Crucial for Nerve Cell Insulation

FROM NIH NEWS, THE NATIONAL INSTITUTES OF HEALTH

Researchers funded by the National Institutes of Health have discovered how a defect in a single master gene disrupts the process by which several genes interact to create myelin, a fatty coating that covers nerve cells and increases the speed and reliability of their electrical signals.

The discovery has implications for understanding disorders of myelin production. These disorders can affect the peripheral nervous system—the nerves outside the brain and spine. These disorders are known collectively as peripheral neuropathies. Peripheral neuropathies can result in numbness, weakness, pain, and impaired movement. They include one of the most common genetically inherited disorders, Charcot-Marie-Tooth disease, which causes progressive muscle wasting.

The myelin sheath that surrounds a nerve cell is analogous to the insulating material that coats an electrical cord or wire, keeping nerve impulses from dissipating, allowing them to travel farther and faster along the length of the nerve cell.

The researchers discovered how a defect in just one copy of the gene, known as early growth response gene 2 (EGR2) affects the normal copy of the gene as well as the functioning of other genes, resulting in peripheral neuropathy.

“The researchers have deci-

phered a key sequence essential to the assembly of myelin,” said Duane Alexander, MD, Director of the NICHD (The National Institute of Child Health and Human Development), the NIH institute that funded the study. “Their discovery will provide important insight into the origins of disorders affecting myelin production.”

The study appears in the online version of *Molecular and Cellular Biology*.

Until this discovery, researchers did not fully understand the complex genetic process that enables Schwann cells, found in the peripheral nervous system, to coat nerves with myelin.

During this study, the scientists found that EGR2 produces a protein that activates several other genes necessary for myelin production. Some of these genes contain the information needed to make peripheral myelin protein-22 (PMP-22) and myelin protein zero (MPZ). MPZ is the most abundant protein in myelin in the peripheral nervous system.

The overproduction or underproduction of the proteins PMP-22 and MPZ account for the majority of inherited peripheral neuropathies, researcher, John Svaren, Ph.D. said.

Ultimately, the sequence of activating genes “switches on” the Schwann cell, which wraps the nerve axon, the arm-like projection that conveys nerve impulses, in a myelin sheath.

The myelin sheath that surrounds a nerve cell is analogous to the insulating material that coats an electrical wire.

The scientists’ research also resolved a long-standing mystery surrounding why a single mutant copy of the EGR2 gene disrupts the functioning of the normal EGR2 gene, leading to a disorder of the nervous system.

In many genetic conditions, the unaffected copy of an affected gene continues to produce its protein. However, the researchers found that the mutant EGR2 copy interferes with the interaction between the normal EGR2 gene and another myelin gene, SOX10, as the two try to work together to produce the myelin protein MPZ.

By understanding the process which creates myelin, researchers may now be able to investigate new therapies for disorders affecting myelin.

“Our research has uncovered a whole new mechanism for regulating myelin genes,” said Dr. Svaren. “Our hope is to exploit this knowledge so that we can adjust the levels of myelin genes such as PMP-22 and MPZ, and thereby create an effective treatment for myelin diseases.” ❄

## The Beron Family and the Creation of Team Julia

BY RACHAEL QUARTNER BERON

I will never forget the feeling I had when Julia was born. I was the most content I had ever been. I had the most wonderful husband, a three-year-old precious son who had survived more in his young life than most (having had successful open-heart surgery at seven months of age), and now a beautiful healthy daughter. Our family was complete. I still feel this way every day.

Life with our two children has not been so easy. That being said, I never felt like my life was harder than anyone else's. Everyone has issues with their children; ours just started earlier than most. It has been hard at times, but I never ever felt like I couldn't handle what cards we have been dealt.

When Julia was six months old, we noticed that her eyes were crossed. We took her to Dr. Caputo for a second opinion

after being told by another doctor that Julia needed surgery right away to correct her lazy eye. He examined her and told us that she had left-eye optic nerve hypoplasia. She had little or no vision in her left eye. Dr. Caputo could fix her "cross-eyed-ness," but her optic nerve was damaged in her left eye and no surgery (as of this time) could correct her vision. Instead, it would be mostly cosmetic. He told us that we should wait until she was 2½ to have surgery, when the rate of success was the best. We had asked him about patching her eyes, and I remember him saying that maybe it would help a little; however, psychologically it could cause damage. Dr. Caputo told us, "Julia's right eye compensates for her left, so let her be who she is." I remember (and live by) those words to this day.

Julia's walking was yet another issue. One year came and went. At first we weren't concerned because I too was a late walker, not starting until 18 months. But soon Julia was 18 months and still no signs. We soon after started her in an early intervention program where she received occupational therapy (OT) and physical therapy (PT). The state of New Jersey provided one session of both OT and PT every week, and we did one session of each on our own. One of her therapists begged me to take her to see Dr. Martin Diamond, a physiatrist at Chil-

dren's Specialized Hospital in Mountainside, N.J. He was the one who after the next 2 years of examining her told us that he thought she had Charcot Marie Tooth. She was tested by him, and then we had the genetic test to confirm his diagnosis. Julia had CMT Type 2E.

We had such mixed emotions at first. My husband was in denial. We had never heard about this until Dr. Diamond mentioned it to us. No one in our family had CMT. Herb and I were tested, and we didn't have any signs. However, many things started making sense to me: wondering why she was falling all the time, why her gait was different from that of her friends, and why she didn't take her first steps until way over two years of age. Now we had a name for this disorder.

When I first learned of Julia's diagnosis, I called my best friend crying. I felt that maybe I had pushed Julia too much over the past couple of years. I remember her saying to me, "Don't you dare stop doing what you're doing with Julia; she is who she is because of what you and Herb have done. Don't let this disease become her."

And we haven't. Julia is a well adjusted, happy, very social, loved, extremely confident, beautiful 7-year-old girl who happens to have Charcot-Marie-Tooth. She is aware of her limitations and is not ashamed of

The Beron family emailed over 500 friends and family to raise research funds.



them. Recently, after meeting with an orthopedic surgeon (who mentioned surgery to Julia), she became very upset. I explained to Julia that this doctor could fix her feet so she could walk straighter. She yelled back at me, "I am different and that is okay. I don't need to walk any straighter. I am fine just as I am!" Enough said about how Julia feels about herself!

A few months ago, Herb and I were talking about how we wanted to start donating to a CMT cause. After doing research on the Internet, I had learned about Steve O'Donnell's swim challenges at the Chesapeake

Bay Bridge. I called him and asked if I could swim with him. He was awesome, inviting me to join him right way. A friend of ours in town, Steve Grossman, offered to swim with me. Herb immediately began working on a massive email/Internet campaign to spread the word (and obtain sponsors) for the swim. The CMT Association offered to construct a website about Julia. That is how Team Julia began.

This has been an extremely emotional time for us. We sent out the e-mail to more than 500 people. Family, friends, clients, teachers, and just about every-

one we knew. The money has poured in, and the support we have been given has been overwhelming. I walk around my town wanting to hug everyone. I am speechless at times, just in awe of how much people care.

So, on June 10th, I am putting on my rented wetsuit, getting in the water at the Chesapeake Bay, and swimming one mile to raise funds for research (to hopefully one day lead to a cure for CMT!). I will be smiling and thinking about how lucky I am to have such a wonderful family and friends that love and care about us so much. I am truly blessed. \*

## As of May 15th, the following people have given generously in honor of Julia Beron:

Mrs. Meredith L. Alin	Mrs. Renee Chiocciariello	Mr. Bill Henderson	Mr. & Mrs. Seth Mendelson	Mr. & Mrs. Jeff Shupack
Mr. & Mrs. Steve Alper	Mr. & Mrs. Lisa O. Chiodi	Mr. & Mrs. Marvin Hochman	Mr. Andy Merkin	Mr. Scott Sklar
Mr. Robert Altman	Coffee Holding Company	Mr. & Mrs. Brad Horowitz	Ms. Jennifer D. Miller	Mr. & Mrs. David E. Slavitt
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Mr. Leslie S. Ash	Mr. Robert D. Correll	Mr. Roland A. Jacobus III	Mr. Jeffrey S. Neubauer	Mr. & Mrs. Michael Sonnabend
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Mr. Fred H. Barrows	Dr. Martin Diamond	Mr. & Mrs. Hyman F. Kleinman	Dr. Robert J. Pearlstein	Mr. & Mrs. Mitchell Storch
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## 26.2 Miles: Going the Distance with CMT

BY STEVE WITT

**O**n January 14, 2007, I was standing in my starting corral, crowded by strangers who would soon be running and walking the P. F. Chang's Rock 'n' Roll Arizona Marathon. It was 6:30 AM and the temperature was around 30° as dawn was approaching. People paced back and forth, jumped up and down, and stepped in place to try and stay warm. The temperature was unseasonably low for Phoenix.

Multiple thoughts, emotions, and reflections flowed through my mind. I felt both excited and energized, and yet strangely calm, in spite of a level of uncertainty of what lay ahead.

**Steve Witt pushed onward to complete a full marathon.**



I zipped up my jacket and pulled the hood up over my head to try and stay warm. “Steve,” I heard Linda, my wife of 25 years, call from outside the corral, “Smile!” as she snapped a few pictures of me.

In less than 3 weeks, I would celebrate my 50th birthday. Today I would walk a full marathon for the first time in my life. 13 years prior I had begun to experience a strange set of symptoms that would progressively affect my balance and stability, making it challenging to stand and walk. My manual dexterity was also affected.

Years had passed with frustration and concern before I received a diagnosis of CMT Type 2 in 1998. I wanted to know what type of treatment was available to help my condition and what I could do to slow the progression. Other than a suggestion to obtain AFOs to stabilize my feet and ankles, I did not find any answers.

I obtained a pair of plastic AFOs and tried them before throwing them in my closet, where they would stay for the next few years. My CMT finally reached a level that forced me to dig the AFOs out of the closet sometime around 2001.

“There has to be something better than these AFOs,” I repeatedly thought to myself. As my mobility declined and my lifestyle became less active, I began an intensive search for an AFO that would enable me to pursue a more active lifestyle,

regain a higher level of mobility, exercise, and improve my level of physical fitness and general health.

In late January 2005, my search would lead me clear across the USA from Virginia to spend a week in Las Vegas, Nevada, with Mitch Warner of Ortho Rehab Designs. On February 3rd, I walked out of the office wearing custom-made Helios AFOs.

I began a very slow and gradual process of walking for a certain amount of time each day in order to get accustomed to walking with my Helios AFOs. As weeks and months passed, my gait became steadier and faster, and I made small but steady progress in my level of physical fitness.

By May of 2005, I started logging the distance and time of my walks in a spreadsheet system that would track my monthly and yearly mileages, as well as pace. I also made a decision that I would accompany Linda to the Virginia Beach Rock 'n' Roll Half Marathon (13.1 miles) on Labor Day weekend. Linda would run in the event and I would attempt to walk, the half marathon within the 4-hour time limit. This goal, which I had registered in my mind as a possibility, became a reality as I completed that event in 3:34:07.

In 2006, I would complete 3 more half marathons, with my fastest time being 3:22:59 at the Virginia Beach event. Plans were

made to walk the full marathon in Phoenix, and I laid out a 16-week training schedule.

I heard the crowd cheer as an announcement was made that the marathon would soon begin. Suddenly, I snapped back to the present moment as a feeling of excitement stirred deep in the center of my being. “I love you!” Linda called to me as she waved and smiled. I looked to the sky and offered a silent prayer of thanksgiving for having been allowed to reach this point. I asked for strength to make it to the finish line in the allotted time.

The gun sounded to start the race and more than 10,000 marathoners began moving. It took almost 6 minutes to get from my corral to the start line where the timing chip tied on my shoe would be activated to start my personal “chip” time.

I had to stop a couple of times before the halfway point for bathroom breaks. One of these was in a church that had offered their restrooms to the marathoners. The multiple layers and my cold hands, which were not working very well, made these stops take longer than usual. I reached the halfway point well under the specified “cut off” time and was allowed to continue on for the second half toward the finish line. I was, however, a good 7 minutes or more behind where I had hoped to be at that point. Yet, I had passed the 13.1-mile (half marathon) point. In my previous events, that had been the finish line, but not today! Today, I still had another 13 miles to go.

Linda was navigating around the area in a rental car

with a map, trying to meet me at various points along the 26.2-mile course to offer encouragement, food, and drink. She managed to see me about six times during the race.

Along the way, crowds would cheer and yell out encouragement. As with all Rock ‘n’ Roll marathon events, bands were performing at intervals along the course. As I approached and passed each stage, the music provided a boost

I asked for strength  
to make it  
to the finish line  
in the allotted time.

of emotional energy which I was finding to be helpful, but too short-lived.

Somewhere before mile 18, I began to struggle mentally and physically. My right knee and thigh began to hurt and my energy level and mental focus began to run low. I had not taken in adequate nutrition and liquid for this long walk. As I passed the support stations, I asked, “Do you have any more goo?” (packets of energy food), but they had run out.

Continuing on was becoming more and more of a “mind game” as I heard a voice in my head say, “You are not going to finish this, so just call Linda to come and pick you up.”

“Don’t listen to that,” I heard another voice say. “You’ve come too far to stop now. Just keep taking one step at a time.”

I came up behind a middle-

aged woman who seemed to be slowing down, but nevertheless, continuing on. She had a message written on the back of her t-shirt. It read, “In 1997, I suffered a stroke ... In 2007, I am doing a marathon.” As I walked up beside her, I put my hand on her shoulder for just a moment and said, “You are doing great. Keep it up.”

Linda met up with me about three quarters of a mile before the finish line to walk with me. This last short stretch seemed like it would not end. Linda continued to encourage me on. “You are almost there,” she would say. “You’ve done it.”

I entered the finish chute, which was fenced off, and Linda stayed along the outside. Less than a hundred yards from the finish line, my legs were cramping up, and I felt as if I couldn’t control them anymore. I was losing my balance, about to stumble and fall. A camera man came to offer me help. I was able to make my way to the fence for some support and to try to summon up enough strength to finish. For a moment I really didn’t know if I could take one more step.

I remember hearing the words in my altered conscience, “Help me Jesus.”

I stepped back onto the course and said, “I’m just going to take little steps.”

As I approached the finish line, I heard the announcer say, “Steve Witt from Christiansburg, VA.” Right after that I heard the sweet chirping sound of the timing chip on my shoe as I crossed the finish line.

I had finished a full marathon, 26.2 miles! ✨

# On the Development Front

BY DAN MERCHANT, CHIEF DEVELOPMENT OFFICER

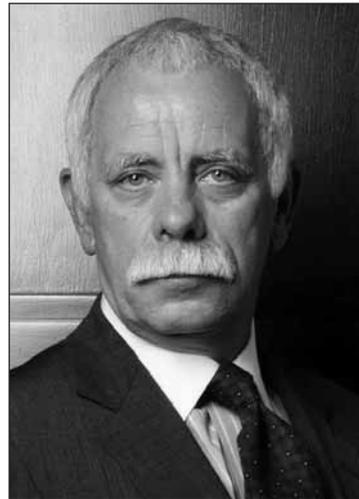
Let me introduce myself and share my personal background and the circumstances that led me to take on the job of raising money needed for the CMTA to achieve its ultimate goal of finding a cure within the next decade.

I came to the CMTA after a career in marketing. A Penn State graduate, I began my career at General Foods in 1969. I spent nearly 20 years running the marketing research and consulting division of Marketing Corporation of America. Most recently I was Chief Marketing officer for U.S. Trust, one of America's oldest and most respected wealth management firms. I have been married for nearly 35 years to my wife, Ann, and have a daughter, Aimée, who is 27 years old and lives in New York City. I have moved from Westport, Connecticut, to Johnstown, Pennsylvania, so that I could work closely with the executive team of the CMTA.

The opportunity to work with this organization and to raise funds is highly motivating and gives me a chance to focus my experience, energy, and enthusiasm on giving back and making a difference. Like far too many people, I learned about CMT only recently. I grew up in Western Pennsylvania and have come back the first Monday following Labor Day to play golf in an annual tournament at Indian Lake Golf Club in Central City, PA, for the past twelve years. It

was at this golf tournament that I met Pat Torchia, Chairman and President of the CMTA. He was introduced to me by Charles Hagins, a long-time friend and executive director of the CMTA.

While at U.S. Trust, I attended a CMTA function in Palm Beach in February 2005 and played my first round of golf with Pat. The course was wet, so



**New chief development officer Dan Merchant hopes to raise \$10 million in two to three years.**

the “cart paths only” rule was in effect. That meant that Pat had to mostly walk the nearly 7,000-yard course. Over the four-plus hours it took to play the round, I heard Pat’s story and observed first-hand the extraordinary courage it took for someone with CMT to participate in an otherwise ordinary activity.

My goal is to raise \$10 million over the next two to three years to fund the research needed to find a treatment and

cure for CMT. So, how do I intend to achieve this goal? Well, it won’t be easy, but I believe that the ground work has already been laid through the arduous efforts of all of those who have worked so diligently since the founding of the CMTA in 1983.

A lifetime of experience in corporate America and event marketing are two assets I bring to the CMTA. Marketing is about fulfilling a need, creating awareness of that need, and communicating its benefits in a compelling way. At the heart it is about selling an idea. Successful marketers find a “unique selling proposition” or USP and a creative way to communicate that proposition to a target audience. The tools available to marketers to reach the audience have expanded and been refined over the years.

The “USP” for CMT is that a cure is within grasp. The genetic nature of the disorder has long been a curse, passed from generation to generation. With the advances in genetic research, that curse has become a blessing. The research that is either in place or in planning bodes well for those who suffer from CMT. I believe there’s an audience out there that will be receptive to helping cure this debilitating and degenerating disorder.

Penn State’s highly successful coach, Joe Paterno, as a CMTA spokesperson presents a creative way to reach a broader

audience with a voice of authority. Penn State has more than 400,000 graduates and the largest alumni association of any university in America. And, of course, you don't have to be a Penn State graduate to know or admire the accomplishments of this football icon. He's more than just one of the most successful coaches in NCAA I-A football history; he is a humanitarian and philanthropist.

"Help Joe Paterno Tackle CMT" is a powerful idea. Football has become America's game.

And, Penn State is a storied team. I hope to leverage this idea and our contacts within the Penn State community to create new initiatives to raise money.

I'm also planning a series of events to attract high-net-worth individuals to the CMT cause. The first is a golf outing in Scotland at Loch Lomond where I am a member. Loch Lomond is the site of the Scottish Open and is rated one of the top 50 golf courses in the world. The club has agreed to donate a three-day event to be held this

coming August that will raise over \$100,000. Other similar events are in the planning stage.

More effective use of the Internet, adding corporate sponsorships, and expanding grassroots events are other vehicles I see as initiatives to help reach the \$10 million goal.

Of course, I cannot do this alone. I will need the help of everyone in the CMTA community to be successful. So, help me help you. Send me your ideas, volunteer if you can. We are long on vision and short on help. ✨

## CMT in the News

*Newspapers around the country feature stories about people with CMT*

Former Philadelphia 76er, Todd MacCulloch, has taken on a new "sport" using his fingers to pursue his passion of playing pinball. MacCulloch bought his first three pinball machines when he played with the New Jersey Nets in 2001-2002. He is currently the 208th ranked pinball player in the world and owns over 50 pinball machines to help him hone his skills.

MacCulloch was forced into retirement by Charcot-Marie-Tooth disorder when the running and jumping demanded of basketball players became impossible. When he isn't playing Medieval Madness or Indiana Jones, Todd is currently doing radio and TV work for the Sixers' broadcast system.

The 10th Annual CMT Daffodils for Seniors took place in April at Laurel View Village in Davidsville, PA. Each spring local

children pick, arrange, and hand out thousands of daffodils to the elderly residents. The daffodil giveaway is in the name of Marah Griffith, a CMT patient who died at age 16 on Christmas Day, 2001. Marah was instrumental in the conception of children giving daffodils to seniors. She loved organizing the event and working with the children, and she particularly enjoyed the pleasure it gave to the nursing home residents.

The Chatham Courier published a story about Bruce Rex, who was first diagnosed with CMT in his twenties, but has gone on to spend much of his life working for the American Red Cross, first as a certified CPR practitioner and then, when his CMT made that difficult, he began teaching CPR to firefighters, college students, and boy Scouts.

Although Rex is retired from his job with CIBA-Geigy

Pharmaceutical Company, he continues to work tirelessly teaching courses on health and safety for his Red Cross chapter, being a hospice volunteer and Past President of his AARP chapter. ✨

**Teenagers delivered daffodils during the 10th Annual "CMT Daffodils for Seniors" giveaway.**



# Wayne State Clinicians Answer CMT Questions

*This is the first of a two-part article by doctors from the Wayne State Clinic. Questions 1-5 are answered by neurologist Dr. Michael Shy; questions 6-10 are answered by neurologist Dr. Richard Lewis.*

## 1 There are so many neurological disorders. How do you determine if a patient has CMT?

CMT, or Charcot-Marie-Tooth disease, requires that the patient have a peripheral neuropathy that is heritable, in other words that can be genetically transmitted. The physician needs to first determine whether the patient has a peripheral neuropathy by taking a history and performing a neurological exam. The physician then needs to determine whether there is a family history of neuropathy. If so, this meets the criteria to call the neuropathy CMT. The next task is to determine which of the many types of CMT the patient has. If there is no obvious family history, the physician needs to determine whether there are features of the neuropathy which suggest CMT. For example, were there features of the neuropathy which started in childhood and slowly progressed or are there features of the EMG or nerve conduction that are typical for CMT and rare for acquired forms of peripheral neuropathy. If these criteria are met, then the specific form of CMT needs to be investigated.

## 2 What physical tests do you perform to validate the diagnosis of CMT?

The diagnosis is based on the history, neurological exam and EMG/NCV. Genetic testing is often not necessary to tell whether a person has CMT or even CMT1 or CMT2. Genetic testing is necessary to determine the specific subtype of CMT a patient has (CMT1A, CMT2A, etc.) because these subtypes are based on specific genes. In tricky cases genetic testing may be necessary to confirm that a patient has CMT (no family history, unclear patterns of NCV, for example).

## 3 If my blood test for CMT is negative, does this mean I do not have CMT?

Commercial genetic testing is only available for about 10 of the more than 30 genes known to cause CMT. There remain additional genes that have not yet been identified so that the total number of genes that can cause CMT is more than 50. If commercial genetic testing is negative, it just means that the patient does not have CMT caused by those genes tested. Even if all 10 are negative, it doesn't mean the patient doesn't have CMT; it just means that he doesn't have a type caused by one of those 10 genes.

## 4 Are there currently any treatments for CMT?

There are many treatments for CMT. There are as yet no cures

for any of the types. Treatments typically involve orthotics, bracing, and rehabilitation. Good diet and health habits in general help patients with CMT. Clinical trials for some forms of CMT, for example CMT1A, are now underway.

## 5 How far away are we from finding a cure or an effective therapy for CMT?

When cures will arrive is impossible to accurately predict because they could happen in the near future or could take many years. Fifteen years ago no genetic cause of CMT had been discovered and there were no disease models to test treatments in. Now more than 30 genes are known and many of these are being used to develop treatments. The CMTA is making a huge effort to facilitate these studies. Many scientists are committed to developing treatments and are spending their careers to develop treatments as fast as possible.

## 6 What do EMG and NCV stand for?

EMG stands for electromyography ("myo" meaning muscle). EMG is usually the term we use for the entire study which includes the nerve conduction test and the needle examination. But specifically, EMG is really the term for the examination of the muscles with the needle insertions.

NCV is the abbreviation for nerve conduction velocity which is obtained with the elec-

trical impulses we give during the test. NCS is sometimes used for nerve conduction studies. These include stimulating motor nerves to the muscles and sensory nerves that deliver messages of sensation.

## 7 What is the purpose of having an EMG done?

The purpose of the EMG and NCS is to determine whether people have problems with their nerves or muscles or the connection between them. For people with suspected CMT, the studies can confirm that the disorder is a primary problem of the nerve and not of the muscle. It can then distinguish between nerve disorders of the axon (the wire of the nerve) or the myelin (insulation of the nerve). In short, myelin disorders make nerves conduct very slowly—usually less than 50% of normal (normal motor nerves conduct at 50 meters/second and in CMT-1A they conduct usually at 25 meters/second). When the axon is the major problem, the nerves continue to conduct close to normal, but the number of nerve fibers functioning is reduced and the amplitude of the responses drops.

## 8 What should a patient expect to have happen during the exam? Does it hurt? Does it always involve needles?

The patient should expect to have a series of nerve conduction studies of the arms and legs. Sometimes only one limb will be studied, but most of the time it is at least two. The limbs will need to be exposed to above the knee and to the upper arm so

the patient may be asked to put on a hospital gown. The nerve conduction studies require electrical impulses that are very safe but are uncomfortable for a “split second.” If the patient has a pacemaker or defibrillator, he should let the electromyographer know. Patients with some forms of CMT have nerves that require a bit higher intensity of stimulation. It is not possible to determine how many impulses will be given, but usually it is 3 to 5 at each stimulation site, and there are 2 to 3 sites for each nerve tested. There is no lingering discomfort except for a slight ache that lasts 10 to 15 minutes. Depending on the clinical situation, needle EMG may be performed. However, not all problems require a needle exam. This should be discussed with the examining physician.

The needles are disposable and carry no risk of infection and are otherwise very safe. If the patient is taking a blood thinner, he should inform the examiner. Virtually any muscle can be tested, but usually the muscles examined are in the arm and leg. The needles are very thin (more like stiff wires) but “needles are needles” and piercing the skin pinches. There can be other very brief discomfort when the needle is moved to find the right area of the muscle. There is no lingering discomfort. Occasionally, a small “black and blue” spot can develop.

## 9 How old should my child be before I consider allowing an EMG to be performed on him?

Both the NCS and EMG examinations can be performed

at any age including infancy. The risks are no greater in children than adults. The infant and toddler will need to be restrained and sometimes sedated (lightly). Some electromyographers find it best if the parents leave the room (many children actually are more calm without the parent although some parents’ anxiety about leaving may outweigh this), but others like having the parent with the child. This should be discussed beforehand. You may also wish to inquire about the examiner’s experience in doing the study in kids. Whether the test is needed in very young infants depends on a variety of factors. Sometimes it may be wise to wait a few years. Other times it is helpful and appropriate to do the studies at a very early age. In general, NCS can determine if a child has CMT-1A by age 5, and there are some studies suggesting differences from normal even in newborns.

The interpretation of results in children under 3 is somewhat different from that in older children because normal nerve conduction velocity is slower in infants than adults. By age 3 or 4 they are close to that of an adult.

## 10 What can you tell from the results?

As mentioned above, the studies can differentiate disorders of the peripheral nerve from those of the muscle and can also distinguish axonal neuropathies (CMT-2) from demyelinating neuropathies (CMT-1). There are some findings that can lead to more specific diagnoses. ✱



## GIFTS WERE MADE TO THE CMTA

## IN MEMORY OF

**Cleo Belz**

Bob &amp; Mary Belz

**Carole Chow**

Rose Yuen

**Mona Donovan**

Sharon &amp; Ronald Gentle

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**Margaret Fingerson**

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Kathleen Intoccia

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**Keith Widdop**

Jenny McReynolds

Pete Mueller &amp;

Cathy Paulson

Jan &amp; Bill Warren

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



# Audrey Spencer MacDonald and A CMT Family Genealogy

BY HEATHER MACDONALD

**A**udrey Spencer MacDonald, who recently died before her 95th birthday, was a student at the Rhode Island School of Design during The Great Depression. It was unusual for a female to be in college at that time. Even more unusual was the fact that she was a New England farmer's daughter and, at that time, only city people were students at RISD.

Audrey's rich ancestry in New England goes back to the seventeenth century when her Spencer ancestor was granted land by the King of England in 1677 when East Greenwich (RI) was incorporated. The first English baby born in East Greenwich was a Spencer baby.

The King of England also granted two Vaughn brothers land when East Greenwich was incorporated. As a young child, Audrey remembered she could hear the banging from the blacksmith shop where her grandfather Vaughn was shoeing horses. Both the Spencer and Vaughn homesteads were in the same family for many generations. Audrey was the last Spencer baby to be born on the Spencer homestead before it was sold in the 1920's.

In Audrey's rich history was a mysterious, upsetting tale of something in the family that was called "the Spencer curse." Excerpts from our genealogical records:

Desire, died, 3-1797 at 73. She married James Greene who in his will provided for lame daughters. Desire Slocum was "lame and decrepit" and Desire's sister, Mercy (1752-1800) Slocum was the second wife of Captain John Spencer. Mercy was a cripple. Mercy's children were: Mercy, who had a daughter who never walked; Ebenezer, who had nothing in his history about lameness; John, who was badly crippled.

Because the defect appeared in John Spencer's children and grandchildren, it was called the Spencer curse even though it came from Mercy Slocum, on the maternal side of the family.

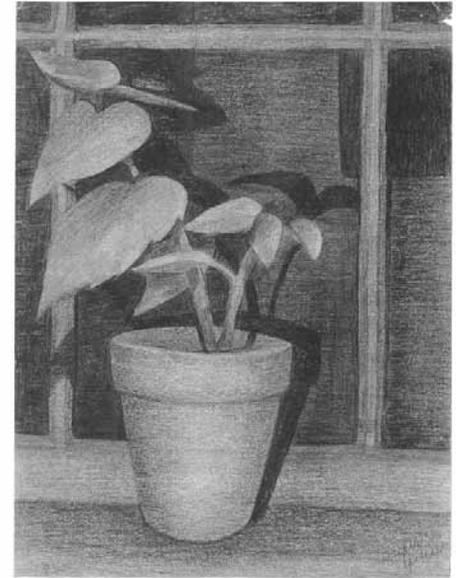
Knowing one's medical genealogy, called pedigree, is very important because I was told that our CMT line was carried on the X chromosome, which means a male has symptoms, but cannot pass the disorder to his sons. He does pass it on to his daughters in the carrier mode. If a female is a carrier, she may or may not show symptoms. Carriers have a 50/50 probability of passing the gene to an offspring. This knowledge gives us the ability to make informed, even enlightened, life choices.

As a child and then as a teenager, when I would overhear my mother and her sister from California quietly talk of the "affliction" in the family, I dismissed it. After all, my three brothers were strong and healthy

and my uncle had become frail and had difficulty walking because of old age—or so I thought. In the 70's when we learned the name for this neuropathy, Charcot-Marie-Tooth, my mother expressed relief because previously this condition had been "swept under the rug." My mother grieved knowing that she had passed this on to her grandson. I told her, "Yes, but you also passed on your good looks, your creativity, your artistic talent, your intelligence, and your inquisitive mind."

Back when I was in my early 20's, I came to California and met my cousins; I realized that this condition was real! This was not something that two sisters made up. To see my California cousins with CMT manage to be successful and to lead quality lives enabled me to see that it is not what you have been given in life, but what you do with what you have been given.

I am grateful to the CMTA for funding CMT research and for educating the public about this most common inherited peripheral neuropathy. Education eliminates fear, brings us wisdom, and gives us the ability to make enlightened choices. \*



**One of Audrey MacDonald's drawings from her days at RISD.**



## SUPPORT GROUP NEWS

### California—San Francisco Bay Area

Dr. Hugh Baras is a psychologist who specializes in Behavioral Medicine and Health Psychology. Cognitive-behavioral therapy is based on the idea that our thoughts cause our feelings and behaviors, not external things, like people, situations, and events. The benefit of this fact is that we can change the way we think so we can feel and act better even if the situation does not change.

Dr. Baras gave us an overview of some cognitive-behavioral techniques he frequently uses in his private practice, which include biofeedback, relaxation, and cognitive behavioral strategies to help people achieve their goals.

He hooked up Mike, one of our brave support group members, to a biofeedback device which displayed hand temperature, heartbeat, arousal level (anxiety), and muscle tension on a large screen for the whole room to observe. By conveying such information to Mike in real-time, he became more aware of his physiological activities and was able to gain conscious control over his own physical processes, which he proved by voluntarily raising his hand temperature, lowering his anxiety levels, and breathing more evenly, smoother, and in a more relaxed fashion.

At the end, Dr. Baras led us through a 5-minute relaxation exercise to show that

everyone is capable of learning to release muscle tension and reduce pain, and anxiety.

The group will hold a family/friends pot-luck picnic on Saturday, June 9, from noon to 5 PM. Call for more details on the location.

### Colorado—Broomfield

Our last meeting was April 28th from 10 to 11:45 AM at the First National Bank. Dr. Dianna Quinn was scheduled to speak, but was unavailable at the last minute. There is exciting news about a future meeting! On June 22<sup>nd</sup>, Dr. Michael Shy, from Wayne State University, has accepted an invitation and will speak to our group. He is a renowned CMT expert and you can read about his clinic at Wayne State in the April/May issue of the newsletter. Please let us know if you will be able to attend, since we need a good attendance to justify having Dr. Shy fly in from Michigan.

### Nevada—Las Vegas

The group will meet on July 21st, at the West Charleston Library, 6301 West Charleston Blvd., in the Conference Room. The meeting will start at 1 p.m. and we are happy to have motivational speaker, Mr. Martin McCorkle, who wrote the book *Walk with Me*.

### Pennsylvania—Johnstown

At our last meeting I was reminded of why support group leaders go through the

often-frustrating exercise of organizing and holding meetings. A new member, the father of a teenager with CMT, drove over 60 miles to attend and was truly appreciative of the information and help he received. New blood is wonderful—it's great to see regular group members, but for the group to really help a new member and try to answer his questions and offer insights into his dilemma is what makes meetings so valuable.

The quest speaker, Tim Golden, an orthotist, discussed the various types and advantages of bracing. A lively discussion debating bracing vs. surgery followed. The guest speaker and co-group leader Jeana Sweeney, who had surgery as a teenager, represented the two sides of the discussion.

### Pennsylvania—Philadelphia

The group met on April 14, 2007, in the community room at 2700 Chestnut St. Member Dennis Devlin arranged for podiatrists, Dr. Nicholas Romansky and Dr. Christopher Corwin, to be available to answer questions from the group members. Topics ranged from how to deal with calluses to how to know what type of orthosis would be most beneficial. It was an informative and lively session with approximately fifteen people in attendance. The next meeting will be on June 16, 2007. ✨

# CMT Support Groups

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

## Alabama—Birmingham

**Place:** Lakeshore Foundation  
Fitness Center

**Meeting:** Call for schedule

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

**Email:** dklrl@yahoo.com

## Arizona—Phoenix Area

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

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

**Contact:** Marilyn Hardy or  
Aisha Hackett, 480-496-4530

## Arkansas—Northwest Area

**Place:** Varies, Call for locations

**Meeting:** Quarterly

*Meetings are not regularly  
scheduled so call ahead.*

**Contact:** Libby Bond, 479-787-6115

**Email:** charnicoma57@yahoo.com

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

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

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

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

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

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

**Place:** Location to be determined

**Meeting:** Bimonthly

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

**Email:** elizabetho@pacbell.net

## Colorado—Broomfield

**Place:** First National Bank

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

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

**Email:** dmcovington@msn.com

## Florida—Tampa Bay Area

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

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

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

**Email:** rathhouse1@verizon.net

## Kentucky/Southern Indiana/ Southern Ohio

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

**Meeting:** Quarterly

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

**Email:** marteye@mis.net

## Minnesota—Benson

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

**Meeting:** Occasionally

**Contact:** Rosemary Mills, 320-567-2156

**Email:** rrmills@fedtel.net

## Minnesota—Twin Cities

**Place:** Call for location

**Meeting:** Quarterly

**Contact:** Maureen Horton, 651-690-2709  
Bill Miller, 763-560-6654

**Email:** mphorton@qwest.net  
wmiller7@msn.com

## Mississippi/Louisiana

**Place:** Baptist Healthplex,  
102 Clinton Parkway,  
Clinton, MS

**Meeting:** Quarterly

**Contact:** Flora Jones, 601-825-2258

**Email:** flojo4@aol.com

## Missouri—St. Louis Area

**Place:** Saint Louis University Hospital

**Meeting:** Quarterly

**Contact:** Carole Haislip, 314-644-1664

**Email:** c.haislip@att.net

## New York—Greater New York

**Place:** NYU Medical Center/  
Rusk Institute, 400 E. 34th St.

**Meeting:** Third Saturday of every other  
month, 1-3 PM

**Contact:** Dr. David Younger,  
212-535-4314,  
Fax 212-535-6392

**Website:** www.cmtnyc.org

**Email:** bwine@acm.org

## New York—Horseheads

**Place:** Horseheads Free Library on  
Main Street, Horseheads, NY

**Meeting:** Quarterly

**Contact:** Angela Piersimoni,  
607-562-8823

## New York (Westchester County)/ Connecticut (Fairfield)

**Place:** Blythedale Hospital

**Meeting:** Bimonthly, Jan, March, May,  
Sept, and Nov; 3rd Saturday

**Contacts:** Beverly Wurzel, 845-783-2815  
Eileen Spell, 201-447-2183

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

## Nevada—Las Vegas

**Place:** Whispering Pines Community  
Center

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

**Contact:** Mary Fatzinger

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

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

**Place:** Various locations in Raleigh

**Meeting:** Quarterly

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

**Email:** judae@bellsouth.net

## Ohio—Greenville

**Place:** Various locations.

**Meeting:** Fourth Thursday,

April–October

**Contact:** Dot Cain, 937-548-3963

**Email:** Greenville-Ohio-CMT@  
woh.rr.com

## Ohio—NW Ohio

**Place:** Medical College of Ohio

**Meeting:** Quarterly

**Contact:** Jay Budde,  
419-445-2123 (evenings)

**Email:** jbudde@fm-bank.com

## Pennsylvania—Johnstown Area

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

**Meeting:** Bimonthly

**Contacts:** J. D. Griffith,  
814-539-2341

Jeana Sweeney,  
814-262-8467

**Email:** jdgriffith@atlanticbb.net,  
cjsweeney@ussco.net

## Pennsylvania—Northwestern Area

**Place:** Blasco Memorial Library

**Meeting:** Call for information

**Contact:** Joyce Steinkamp,  
814-833-8495

**Email:** joyceanns@adelphia.net

## Pennsylvania—Philadelphia Area

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

**Meeting:** Bi-monthly

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

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

## 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



## ASK THE DOCTOR

### Dear Doctor,

*I have CMT. I have been taking a vitamin supplement called L-arginine. I noticed today that it had vitamin B6 10 mg (as pyridoxine HCl). One pill is 500% of the daily value. You can take from one to six tablets per day. I have been taking two to four. Since mega doses of vitamin B6 are listed as something not to be taken as a CMT patient, should I stop taking this? I am taking this on my own accord, as I take a few other herbal supplements recommended in some holistic books.*

### The doctor replies:

The highest safe dose of pyridoxine (vitamin B6) is somewhat controversial, but may be lower than generally accepted. It is one of a small number of drugs that specifically target the same type of nerves affected by CMT. Most known cases reported were taking 200 mg or more a day, but there are reports of possible or probable problems at 100 mg and maybe even 50 mg/day. One or 2 tablets a day is probably safe, but 6 tablets/day starts to stray into the gray area. If there is concern, a blood level of B6 can be measured by some laboratories, but the test is not universally available.

### Dear Doctor,

*I am an ambulatory patient with CMT and have worn leg braces for the past two years. I consider my CMT to be relatively mild, though in the last thirty years my hand and feet*

*have been progressively affected to a greater degree.*

*I have had two operations and a spinal block was suggested each time. Fearing that any intrusion into my spinal cord might possibly cause the CMT to get worse, I elected to have a general anesthesia. I'm wondering if I was overly cautious. Is it possible that the myelin sheath of the spinal cord might be adversely disturbed by a spinal injection?*

### The doctor replies:

The myelin of the brain and spinal cord is different from the myelin of peripheral nerves, but that should not be an issue. It is my understanding that "spinal anesthesia" is usually injected just outside the structure called the dura, and never into the spinal cord itself, so that there is little risk of injuring nerves or nerve roots. I believe that spinal anesthesia is safe for patients with CMT.

### Dear Doctor,

*My Mom has CMT and her doctor gave her Mirapex (another name for it is pramipexole. She wonders if it is safe to take.*

### The doctor replies:

Pramipexole (Mirapex) is an increasingly used medication for both Parkinson disease and restless leg syndrome and for off-label use in certain behavioral situations (compulsive gambling.) There is no known link with CMT or toxic neuropathy; however, restless leg-like symptoms can occur because of neuropathy or from other causes.

Mirapex has side effects unrelated to neuropathy that should be reviewed with your physician before starting the medication.

Another drug pergolide [Permax] that works in a similar way (boosting dopamine) was recently taken off the market because of concerns of damaging heart valves. Mirapex, however, is from a different class of compounds and has not been linked with this complication.

### Dear Doctor,

*My son is 50 years old. He has CMT and also schizophrenia. He is taking Clozaril and Effexor. He also takes Prilosec. Recently, the doctor added an older psychiatric drug, Trilifon. Now, my son is complaining of what he calls "spasms" in his feet. He wears supports for both feet and his hands are also affected. Are any of these medicines contraindicated?*

### The doctor replies:

Psychiatric drugs have a variety of side effects, especially some of the older agents such as Trilifon (perphenazine), which is in the phenothiazine class. Muscle spasms are a common and well-recognized side effect, but one that has little to do with CMT. The psychiatrist should explain these effects and adjust the dose accordingly. Clozaril and Effexor also have side effects but are generally better tolerated. None of these drugs is implicated in worsening neuropathy, however.

### Dear Doctor,

My reproductive endocrinologist is concerned that undergoing In

## WRITE TO US!

Pat Dreibelbis, Editor  
The CMTA Report  
CMTA

2700 Chestnut Pkwy.  
Chester, PA 19013

or

info@charcot-marie-tooth.org

Vitro Fertilization (IVF) and having to use progesterone in the form of shots and suppositories for 8 weeks will have a toxic effect on my CMT, given the fact that progesterone antagonists may be beneficial for CMT patients. Can progesterone cause toxicity or hasten my CMT progression?

### The doctor replies:

The effects of the progesterone blocker are purely investigational in laboratory animals at this point. No positive or negative effect has been noted in humans, so the side effects from IVF on CMT are purely speculative. I know of no information about the effects of hormones related to IVF in any report. However, there is some suggestion that a minority of women have some worsening of weakness during pregnancy, but it is unknown what the degree of change is or what the cause of this effect might be.

### Dear Doctor,

*I was wondering if the new pill ptc124 would help people with CMT. Have any tests been done?*

### The doctor replies:

PTC124 is a new, orally administered small-molecule compound that targets a particular genetic alteration known as a nonsense mutation. Only certain rare types of CMT are caused by this particular type of gene defect. I am not fully up to date on the details, but it appears that the compound is only in the earliest stages of human testing but appears promising. The literature suggests that the first attempts to treat disease will be in patients with cystic fibrosis and Duchenne muscular dystrophy. \*

## IN HER OWN WORDS

I have CMT Type 1. I have only recently been diagnosed with this, even though my problems started at a young age. When I was younger, I found it hard to walk. I didn't walk properly until I was about two years old, and even now I don't really walk normally because my left foot does not allow me to do so. I cannot do the heel-to-toe step like any normal person. My "nan" also noticed when I was younger that when I ran my legs stuck out and I ran on my tip-toes.



It was when I turned twelve or thirteen that my problems really started. I remember walking with friends and suddenly feeling a huge shot of pain soaring through my left foot and leg. The pain lessened toward the end of the day, but it still hurt for two or three weeks after. I thought it was a one-time thing, but about a month after the pain occurred, it was back again. I was on holiday with my friend and her mother, and I was in so much pain that I ended up crying while on the beach. My friend and her mother couldn't understand what was wrong. Like the pain before, this one left after a few weeks. Then, about six weeks later, it started again. This time, it was so painful I couldn't get out of bed and even when I was in bed, it still hurt a lot. My nan took me to the hospital where the doctors put a plaster cast on my leg, even though it wasn't broken, so I am able to walk around to get to school.

My life pretty much went on like that, with the pain going and coming back. My nan got in touch with my doctors and asked them to find out what was going on. So, at fifteen, things started to get moving. I went to see a lot of doctors, because I kept being referred from one to another. Finally last winter, I met an orthopaedic doctor who suspected I had CMT and sent me for electrical tests. After I completed these, he was certain I had it. He is going to operate on my foot and now I see both him and a neurologist who has advised me to do physical therapy. This Easter the pain got a lot worse and it hasn't left, so I am in pain every day. It isn't always as bad, but it's still there, like a 'niggie' pain. Four times since July, I have broken down and cried because the pain is so great. My boyfriend carries me at times like this because I cannot stand up.

I know the pain is going to get worse, so I am just preparing myself for that. I am not going to let it stop me from following my dream of teaching. I am now studying for my A-levels and I am trying hard to catch up on the work I missed because of my CMT. My teachers have been wonderful and are really caring and always ask how I am.

I am sixteen years old and I want the life of a normal teenager, so I do everything my friends do in spite of the pain. My family and friends are really supportive. I don't know what I would do without them.

—Hayley Dixon, United Kingdom

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## CMT PATIENT MEDICATION ALERT:

### Definite high risk (including asymptomatic CMT):

Vinca alkaloids (Vincristine)

### Moderate to significant risk:

Amiodarone (Cordarone)  
Bortezomib (Velcade)  
Cisplatin and Oxaliplatin  
Colchicine (extended use)  
Dapsone  
Didanosine (ddl, Videx)  
Dichloroacetate  
Disulfiram (Antabuse)  
Gold salts  
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 U.S.)  
a-Interferon

### Negligible or doubtful risk:

Allopurinol  
Amitriptyline  
Chloramphenicol  
Chlorprothixene  
Cimetidine  
Clioquinol  
Clofibrate  
Cyclosporin A  
Enalapril  
Fluoroquinolones  
Glutethimide  
Lithium  
Phenelzine  
Propafenone  
Sulfonamides  
Sulfasalazine

# What is CMT?

- ◆ is the most common inherited neuropathy, affecting approximately 150,000 Americans.
- ◆ may become worse if certain neurotoxic drugs are taken.
- ◆ can vary greatly in severity, even within the same family.
- ◆ can, in rare instances, cause severe disability.
- ◆ is also known as peroneal muscular atrophy and hereditary motor sensory neuropathy.
- ◆ is slowly progressive, causing deterioration of peripheral nerves that control sensory information and muscle function of the foot/lower leg and hand/forearm.
- ◆ causes degeneration of peroneal muscles (located on the front of the leg below the knee).
- ◆ does not affect life expectancy.
- ◆ is sometimes surgically treated.
- ◆ causes foot-drop walking gait, foot bone abnormalities, high arches and hammer toes, problems with balance, problems with hand function, occasional lower leg and forearm muscle cramping, loss of some normal reflexes, and scoliosis (curvature of the spine).
- ◆ has no effective treatment, although physical therapy, occupational therapy, and moderate physical activity are beneficial.
- ◆ is usually inherited in an autosomal dominant pattern, which means if one parent has CMT, there is a 50% chance of passing it on to each child.
- ◆ Types 1A, 1B, 1C, 1D (EGR2), 1E, 1F, 1X, 2A, 2E, 2I, 2J, 2K, 4A, 4E, 4F, HNPP, CHN and DSN can now be diagnosed by a blood test.
- ◆ is the focus of significant genetic research, bringing us closer to solving the CMT enigma.

## THE CMTA Report



2700 Chestnut Parkway  
Chester, PA 19013  
1-800-606-CMTA FAX (610) 499-9267  
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