

The CMTA Report

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A resource for information on Charcot-Marie-Tooth disease (Peroneal Muscular Atrophy or Hereditary Motor Sensory Neuropathy), the most common inherited neuropathy.

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A REPORT FROM VANCOUVER

Growing Interest in CMT Research

By **ANN LEE BEYER, Chairman and President**

Over the past 19 years, as part of our outreach program, the CMTA has staffed a booth at a number of neurology, genetics, and other medical meetings. In spite of the fact that CMT has rarely been seen on their agenda, we have continued to maintain a presence at these meetings and disseminate information on CMT and the CMTA.

I remember working at the CMTA booth the first day of the American Neurological Association meeting in Toronto, shortly after our Third International Conference on CMT Disorders, and seeing one of our Medical Advisory Board members heading for our booth. He was smiling and looked quite pleased. The reason he was so happy was that CMT had been mentioned in the opening address. CMT had actually been acknowledged at a neurology meeting! We were making progress.

In the five years since the Third International Conference, with the many advances that are taking place, CMT and the CMTA are getting more attention. We are happy to report that the number of poster and platform presentations on CMT at the American Society of Human Genetics as well as the American Academy of Neurology and American Neurological Association meetings is steadily increasing. So is the number of journal articles.

This past July, we staffed a booth at the Tenth International Congress on Neuromuscular Diseases in Vancouver, Canada. This congress meets in various cities around the world every

five years and we had never had the opportunity to have a booth at this meeting. Because it was being held in North America, and knowledge about CMT is increasing at a rapid rate, members of our Medical Advisory Board thought that we needed to be there.

It was one of the most important and exciting meetings we have ever attended. For the first time, CMT played a significant role at a major neurological meeting. There were over 20 poster presentations on CMT disorders at this congress, and several platform or oral sessions where

CMT was the topic of discussion. CMT is finally getting the attention it deserves.

This meeting brought together CMT researchers from around the world who are looking at CMT from many different perspectives. Some are working with animal models and looking at molecular pathways. Others are molecular biologists searching for biological causes of CMT

through the regulation of gene expression. Others are studying the same group of diseases by focusing on how Schwann cells (which make myelin) and axons interact with each other. Another researcher is trying to understand what actually goes on inside the nerve by examining old biopsies. Interestingly, he has discovered that, sometimes, genetic diagnoses can actually be made from old biopsies.

Presenters came from Australia, the Czech Republic, England, France, Germany, Israel, Japan, Moldova, Portugal, Russia, Saudi Arabia, Scotland, Taiwan, and, of course, the United States. Many of them are conducting cutting-edge

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The CMTA.... Living our Mission

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

Vision Statement:
A world without CMT.

The poster which reported on the North American Database garnered attention at the International Congress in Vancouver.

GROWING INTEREST IN CMT

(Continued from page 1)

research in CMT and their presentations reported many new breakthroughs in the genetics, pathology, and molecular biology of the disorder.

A number of the studies were based on the results of collaborative efforts that are taking place among investigators from different institutions and countries: Algeria and France, the Czech Republic and Germany, Germany, the United States and Israel, Germany and Russia, Israel and Great Britain, Israel and Belgium and Saudi Arabia and France. Collaborative research is also being carried out among researchers from different institutions in the United States.

The more CMT researchers interact with each other and share their knowledge and theories, the more it helps. On the basis of the two international congresses that the CMTA sponsored, we know that these gatherings generate new ideas and new ways of looking at CMT, and this leads to significant breakthroughs. Now, they also lead to more collaborative studies both international and domestic. The more collaborative studies, the larger the numbers of CMT patients that can be evaluated and the more knowledge that will be gained.

It was also gratifying to receive some very positive feedback from a CMT researcher who had attended our Third International Conference. He is from the Czech Republic and told us how impressed he was with our organization and our accomplishments. He was so impressed that he went home and started a similar organization! Researchers from his institution are involved in major CMT research and presented three posters at this meeting.

Our booth was busy and a number of attendees from other countries visited to ask about affiliating with the CMTA and also about our grant application process, so that they might become CMTA fellows. Our continuing presence at these meetings is paying off. We *are making a difference*.

CMT1A, HNPP, CMT1B, CMT2, autosomal recessive CMT2, CMT4A, CMT4E, and CMT5 were all addressed at this meeting.

The session on the CMT North American Database generated a great deal of interest. To date, over 500 forms have been received, and 234 people are now registered. Of these 234, 103 have CMT1A, 69 have CMT2, 37 have CMTX, 17 have HNPP, and 8 have CMT1B. Another 271 people either have not been assigned a genetic type or have not undergone genetic testing. We are happy to report that information from the database is now available for studies by qualified researchers.

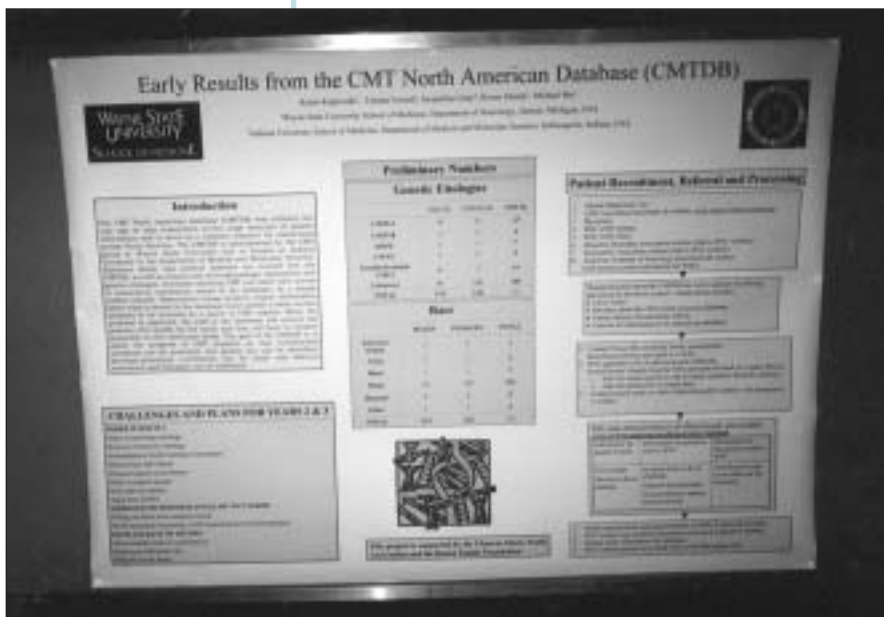
Also of interest was a preliminary report of an ongoing longitudinal study of the most common form of CMT, CMT1A, which measures the natural history of CMT. The purpose of this study is to determine the disease progression that takes place as patients age. As treatments become available, this information will be invaluable.

There were five presentations on HNPP (hereditary neuropathy with liability to pressure palsies). This, too, is a first. Generally, HNPP gets very little attention. One detailed study of the phenotype spectrum of HNPP presented a more complex clinical picture than has been previously reported. A second presentation was an electrophysiological study on lower arms and legs that showed that there seems to be a characteristic pattern on nerve conduction studies that accompanies HNPP.

In addition, there was a very interesting study of families that have HNPP but do not have the deletion of the peripheral myelin protein (PMP22) gene, which is the cause of HNPP. An overproduction of this gene causes CMT1A. Rather, these families have a point mutation in the PMP22 gene—a very rare occurrence.

Peripheral myelin protein (PMP22) took the spotlight again when it was reported that it can cause a significant form of axonal recessive CMT (a T118M homozygous mutation), a recessive form of CMT in which both parents carry the gene but do not have symptoms of the disorder. Axonal forms of CMT are usually classified as CMT2, so this, too, is quite interesting and very surprising.

Other autosomal recessive forms of CMT—which tend to be quite severe—were addressed



at the conference, too. One study identified a new mutation—LMNA—connected with an autosomal recessive form of CMT2, a hitherto unknown form. This is a significant finding.

Finally, there is the peripheral myelin protein zero (MPZ) gene. A mutation in this gene causes CMT1B, generally less severe than CMT1A. However, the MPZ gene has also recently been tied to a severe form of CMT1 and a severe demyelinating infantile-onset form of CMT. To confound things more, it has also been recognized that, clinically, the MPZ mutation can look like CMT2. Investigators are trying to figure out why this MPZ mutation can cause such different clinical associations.

I have included this information to demonstrate that major breakthroughs are taking place, and also to show the complexity of CMT. There are still so many unanswered questions and so much more research to be done before CMT is fully understood.

As stated earlier, our CMT North American Database is functioning very well. Registrations are being processed as they are received. Remember, CMT researchers and clinicians are depending on you to send for and fill out the forms and to register in the database. The information all of you provide will help them gain a much clearer understanding of the full range of symptoms that accompany the various types and subtypes of CMT.

We are receiving registrations for the first annual meeting of the CMT North American Consortium, which is set to meet in March 2003. The consortium is certainly going to stimulate new ways of looking at CMT that will lead to breakthroughs and generate more collaborative studies, especially since our investigators will be meeting with their European counterparts every three years.

Think about the wealth of information that is going to become available to CMT researchers and clinicians from our database and consortium projects and the possibilities that this knowledge will present.

Finally, so far this year, we have received six proposals for our 2003 \$35,000 postdoctoral grants, and we know of one more that is on its way. All are from CMT investigators in top institutions. If they all receive good ratings from our Grant Review Committee comprised of their peers on our Medical Advisory Board, it will cost \$245,000 to fund these one-year studies. We hope that we will be able to fund them all!

The more we learn about CMT, the closer we are to better treatments and, finally, a cure.

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Name: _____

Address: _____

Phone Number: _____ Email: _____

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

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Contribution to CMT Research <i>10% will be applied to administrative expenses.</i>			
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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):

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Professionals Working for CMT Patients: Dr. Gareth Parry

At present I am Professor of Neurology at the University of Minnesota in Minneapolis, having recently returned from a leave of absence in Auckland, New Zealand. I am Director of the Neuromuscular Program and Medical Director of the Clinical Neuroscience Research Unit.

I grew up in New Zealand and, after a brief flirtation with a career as a musician with the New Zealand Symphony Orchestra, I enrolled in medical school in 1966. I had no great interest in the neurosciences during medical school, initially anticipating entering primary care or possibly pursuing a career in cardiology. During my final year at medical school, I had the privilege of working with Dr. Martin Pollock, an expert in peripheral nerve disorders who had recently returned to New Zealand from the United States, and he stimulated my interest in neurology as a career.

My interest in neuropathies began in my early postgraduate years in New Zealand when I saw a teenager who was referred with a diagnosis of CMT but who turned out to have an acquired neuropathy called chronic inflammatory demyelinating polyneuropathy (CIDP). Dr. Pollock asked me to present the patient at grand rounds and I began to read about CMT and CIDP. This launched a career-long interest in neuropathies in general and CMT in particular. In New Zealand, and in many other countries, neurology is considered a branch of internal medicine, so I completed my medicine residency but maintained close contact with neurology. In 1976 I began my neurology residency at the University of Pennsylvania, where I worked under several influential peripheral nerve experts including Drs. Arthur Asbury, Austin Sumner, and Mark Brown, further cementing my interest in neuropathies. One of my fellow residents in the training program at Penn was Dr. Richard

Lewis, another current member of the CMTA Medical Advisory Board. After stints on the faculty of the University of Pennsylvania, the University of California, San Francisco, Hahnemann University in Philadelphia, and Louisiana State University, I joined the faculty at the University of Minnesota in 1992 and have remained here apart from my recent leave back in New Zealand.

Although, as mentioned above, I had been interested in CMT since my early postgraduate years, my fascination with these disorders was further enhanced when I was working in New Orleans. CMT1A is very common in the Cajun population of southern Louisiana and many of these patients were followed by Dr. Carlos Garcia at LSU. He was working with Dr. Jim Lupski from Houston to isolate the gene responsible for the disorder. I began working with Dr. Garcia around the time the gene was isolated and was fortunate enough to go on several field trips into Cajun country to see these patients in their homes. I recall one particularly large family who



Dr. Gareth Parry

arranged a crawfish boil and invited over 100 relatives from near and far to join them. Once they were there and enjoying themselves, the matriarch of the family then announced that the doctors from LSU were there to examine everyone, including doing nerve conduction studies, that were done with the subjects lying on the kitchen table. Studies done in members of this family, along with smaller numbers from several other families, provided the data that allowed us to confirm the original observations of Drs. Lewis and Sumner that the pattern of nerve conduction abnormalities in CMT1A was absolutely consistent and enabled a reliable distinction to be made between CMT1A and CIDP. If I had known about this many years earlier, I would have experienced much less difficulty in recognizing

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Respiratory Issues in CMT: A Personal Story

By J. D. GRIFFITH

MEMBERSHIP RENEWALS

Membership renewals are sent six weeks before the actual expiration of your membership. When you pay your dues, your account is considered current for one calendar year from the date of your renewal.

Frances Gunter's ex-husband, John Gunter, wrote the now classic memoir, *Death Be Not Proud*, after their son Johnny died in 1947, at age 17, from a brain tumor. In it Frances writes of survivor's guilt:

"Missing him now, I am haunted by my own shortcomings, how often I failed him. I think every parent must have a sense of failure, even of sin, merely in remaining alive after the death of a child. One feels that it is not right to live when one's child has died, that one should somehow have found a way to give one's life to save his life. Failing there, one's failures during his too brief life seem all the harder to bear and forgive."

My 16-year-old daughter, Marah, died of respiratory failure on Christmas Day, 2001, in Vail, Colorado, while visiting her aunt and uncle. She had been diagnosed with Charcot-Marie-Tooth (CMT) type 2 phenotype and needed AFOs. On Christmas morning, she called home and said she "couldn't breathe" and was using her cousin's nebulizer (Ventolin, for asthma) when she subsequently collapsed.

I believe it is time that the CMTA remove the phrase "Does not affect life expectancy" from the "What is CMT?" description in the literature and website.

Dr. Ben Galloway, a Denver forensic pathologist, in his autopsy report listed the cause of death as "respiratory failure due to, or as a consequence of, Charcot-Marie-Tooth Disease." Dr. Galloway and his staff spent an extraordinary amount of time and effort researching CMT. Dr. Galloway's autopsy report and protocol were reviewed by the pathology department of the National Jewish Hospital in Denver, CO. This hospital has been ranked number one in the nation for excellence in treating respiratory diseases and first in reputation, among pulmonary specialists, four years in a row by *U.S. News and World Report's* "America's Best Hospitals."

I joined the CMTA in 2001. I received their literature and explored their website, which contained no information on breathing problems. CMTA is not alone. In an informal survey of U.S. websites describing CMT, only Yale's Department of Neurology mentions diaphragm weakness, probably as a result of Dr. Charles

Chan's work. I found a University of Antwerp (Belgium) CMT questionnaire that includes diaphragmatic dysfunction and vocal cord paralysis as two of the 12 questions concerning symptoms. CMT, in the United States, is not considered life-threatening. Do Europeans know something that we don't?

Following my daughter's death, I spent time researching CMT and the involvement of CMT with respiratory failure. I contacted the Canadian group (no longer active) and spoke with its director, Linda Crabtree. She has severe breathing difficulties and vocal cord problems as a result of her CMT. Linda sent me information on the involvement of CMT and the phrenic nerve (innervates the diaphragm) and vocal cord dysfunction (can mimic asthma and restrict air flow). Linda also sent me a disturbing email suggesting that if I had access to this information earlier, my daughter's death might have been prevented.

While reflecting on my family, I realized that not only do we have CMT symptoms (pes cavus, hammer toes, carpal tunnel problems, and peripheral muscle weakness), but we also have a history of respiratory problems and sudden death. My sister (55) went into respiratory arrest in 1990 while in the hospital for a colonoscopy. She was told, had she not been in the hospital, she would have died. She said it felt like an elephant was on her chest. Electrolyte imbalance, as a result of bowel cleansing, was suggested as a possible cause.

I (58) made two visits to the ER with severe breathing problems two years after I fractured a collarbone and some ribs while skiing. My mother and brother died suddenly. Their death certificates listed other causes; however, no autopsies were performed. In hindsight, the cause might have been respiratory arrest.

The Color of Me

Blue is the song of my heart
My heart and soul are often blue
Descending downward
Two eyes weeping blue tears
Representing oceans of emotions throughout the years
Tranquil moments in time
Singing my life's rhyme

—Marah Griffith

My mother, sister, and I have all been diagnosed with asthma and have used inhalers.

Numerous citations exist in the literature concerning the involvement of CMT with phrenic nerve and vocal cord problems. In an article in *The Archives of Internal Medicine*, Dr. Chan et al. state, “based on evidence in the literature and in this field study, a significant number of CMT patients will go on to develop respiratory weakness.” Dr. Greg Carter et al., in a study reported in *Muscle and Nerve*, found abnormalities in phrenic nerve conduction in 22 of 23 patients when a response was obtained. In a conversation with Dr. Carter, he told me that “the phrenic nerve is generally more affected than the peripheral nerves” in CMT patients. He also stressed that “CMT and breathing problems should be further investigated; however, no funding is available. I say ‘further’ only because we know that breathing is involved and impaired in CMT from prior work done by Dr. Chan and myself and others.”

Why isn't more research being done on the problem? It is certainly not for lack of urgency, but for lack of funding!

CMT and serious respiratory problems may be unique to my family's genotype, but I doubt it. I recently discovered an older CMT patient with severe CMT-related breathing problems in my small town. If you have CMT and are experiencing breathing problems, you should consider it a deadly serious matter.

CMT is a diabolical disease. The sinister progressive impairment of lifestyle it generates in adults pales in comparison to the incremental devastation that CMT wreaks on our children. My heartache, as I saw the courage of my beautiful teenage daughter as she struggled to maintain the illusion that she was normal, while expressing her torment through her poetry, was indescribable. My vain attempts to share her pain, such as when she was blown over into a mud puddle in front of her classmates, was unheeded.

My belated awareness of CMT-related breathing problems opened the door for me to connect CMT with respiratory failure in my family, and I hope it will do the same for others. If anyone knows of people with similar situations, please contact the CMTA or me at jdgriffith@mail.charter.net or J. D. Griffith, 3013 Old Somerset Pike, Johnstown, PA 15905.

Please, let's all get together and raise awareness and money to fight CMT. Start a support group, organize a fundraiser, or buy a CMTA golf shirt and wear it (be prepared for some odd questions). Believe me, one person can make a difference. Just look at what the CMTA Board

Come Take a Walk in my Shoes for a little While

Down the crowded street I walk trying to stand tall
People look down and stare at me
I know exactly what they are staring at
It hurts me
It's like an arrow piercing my heart
If I could change it I would
With the blink of the eye I would change it
But I can't
For some strange reason God has dealt me a strange
and painful hand
Why is it that some people seem to float right through
the crooked path of life without any pain or trouble
When me or others have so much pain on our
worried hearts
It does not make any sense to me
It's all a mystery to me
I can't sit and wonder why I have this pain
and others don't
But instead I have to wake up each morning
and try as hard as I can to be as strong as steel
And know that in the end I will be the stronger and
wiser person
since I'm the one that has to deal with this horrible
pain in my heart
Never letting others know how much I really do hurt
inside
I wish people understood
but
they can't
Their minds can't comprehend or even know the pain
that some of us have to walk with every day
Our worries are so much bigger than theirs
It's not their fault they can't understand our pain
But they should be careful not to be so quick to judge
because that person they are judging could be a truly
wonderful person full of love and life
Always ready to share it
So, next time don't laugh and gawk
Maybe think about what it is like to be them
Why don't you take a walk in my (or their) shoes for a
little while?

—*Marah Griffith*

members have done for CMT. If you inspire two people and they each excite two people, now nine people are making a difference. Seven more iterations and 20,000 enthusiastic supporters will make a difference.

It will take some of your time and energy, but to save our children, future generations, and just maybe us from this curse, this is a small price to pay. To paraphrase Barry Goldwater's 1964 presidential nomination acceptance speech, “extremism in the defense of ‘our children’ is no vice. And let me also remind you that moderation in the pursuit of ‘this goal’ is no virtue.”

Contact the CMTA for ideas and help. Together we can de-*feet* this insidious disease.

Three Fundraising Events Support CMT Research

The latest total on the Steve O'Donnell Swim for a Cure is \$69,901. Congratulations on a spectacular job!

On July 22, 2002, the exclusive Creek Club in Locust Valley, New York was the venue for the Third Annual AFA Protective Systems, Inc. Golf Outing for the benefit of the CMTA.

This year's event attracted a full but intentionally limited field of 72 golfers, who enjoyed a fast-paced round on a historic golf course ranked in the top 100 courses in the country.

A bevy of prizes were awarded after play, highlighted by a Cartier watch won in a shootout after the round by Ed Rosenbaum by a margin of three inches. Team prizes, including Waterford Crystal and merchandise from the Creek's Pro Shop, were awarded to half of the entire field, as well as prizes for closest to the pin, longest drive, and straightest drive. The day also saw Frank Briguglia make a hole-in-one on the fourth hole for which he received two free airline tickets.

In the end, not only did everyone have a great time, but the event raised over \$40,000, after expenses, to be used for CMT research.

We particularly would like to mention the many underwriters and donors that helped make the event so successful. AFA Protective Systems, Inc., the oldest central-station alarm company in the country, with offices throughout the East Coast, underwrote the bulk of the cost of the event. Sanders, Sanders, Block & Woycik, a law firm located on Long Island, once again supported the event with many participants and extra funding. Other entities that supplied prizes or giveaways for participants included Bryant & Cooper Steakhouse, J. P. Morgan Chase, Lindt & Sprungii, European American Bank, First Data, WLNY-TV, Land Rover of Glen Cove and Yankee Candle. The CMTA recognizes that the success of the event was largely dependent upon these entities and we appreciate their support and participation.

—Robert Kleinman



Right: Pamela Kleinman and Sheila Sharpe, seated, were joined by Dick Sharpe and Steve O'Donnell at the registration desk for the Creek Club golf outing.



The Second Annual CMTA Golf Outing at Sunnehanna Country Club in Johnstown, PA took place on July 26, 2002. Held in memory of Marah Griffith, the outing was a huge success in spite of a minor rain delay. CMTA Board member Patrick Torchia and Executive Director Charles Hagins worked to organize the event, which raised over \$15,000 for CMT research.

The golfers were delighted to play one of the finest golf courses in America and, at the same time, contribute to the cause of finding a cure for CMT. The competition was fierce but friendly, and some lighthearted banter about “ringers” and “inflated handicaps” added to the fun of the afternoon.

A new addition to the festivities was a silent auction, which followed a cocktail party and dinner. A plaque honoring the poetry of Marah Griffith, who died last year from complications of CMT at age 16, was presented to her father, J. D. Griffith, by golf participant, William Polacek and other local business leaders.

—J. D. Griffith

A third golf tournament is scheduled for October 1, 2002, at the Radnor Valley Country Club in Radnor, PA. This Second Annual John J. Scarduzio Memorial Golf Outing recognizes the 26th anniversary of the death of organizer Christopher Scarduzio's father. His father suffered from CMT and he will dedicate the proceeds from this golf outing to funding CMT research so others can benefit from the ultimate cure of the disease. Last year's golf tournament netted \$18,775 for the research fund.

YOU Can Make a Difference!

What can one person do to help the cause?

■ **Send for the database form.** Fill it out as completely as possible and mail it back to Wayne State. Yes, it is long and takes time to do. As you know, CMT is a complex and still much misunderstood disorder. The information you provide will be invaluable in helping clinicians and researchers define the range of symptoms that comprise CMT. This will not only speed up the pace of research, but it will also help us get better medical care.

If you have questions, or need help, there is a research assistant at Wayne State, Raven Lewis Martin, who is available to walk you through the process. Her number is 313-577-5273. Think of filling out the form as an investment in yourself and your family.

A little time and some postage...that's all it takes to make a difference.

■ **Get involved in a CMT research project.** The CMT Clinic, at the Neurology Department of Wayne State University in Detroit, is looking for patients to enroll in their CMT research studies. They are doing research on all types of CMT.

The center is the only one of its type in the country and the neurological exam will be the most thorough exam you have ever had. In addition to improving patient care, information gathered from patient evaluations is being used for research purposes to learn more about CMT.

They see patients from all over the country and there are affordable rooms where you can stay right on the campus. For more information, call Lisa at 313-677-1689 or check out the clinic website at www.med.wayne.edu/neurology/cmt.html

■ **Start a CMT support group.** We have a written, step-by-step guide to help you start one, as well as a support group leader and an office staff who will be there to guide you through the process and answer any questions you may have.

■ **Do a fundraiser for CMT research.** Golf tournaments have been quite successful, and we can provide guidelines to help you.

■ **Contribute to our CMTA Research Fund.** Whatever you can afford to give will help us to ultimately help you. If you can afford to fund a postdoctoral CMT research fellow for one year, that would be fantastic!

ISSUES AFFECTING CHILDREN

Scoliosis... A Secondary Concern with CMT

The healthy human spine curves in a gentle "s" from back to front, but in some youngsters, the spine also curves abnormally from side to side. This is the defining characteristic of scoliosis.

For reasons that are unknown, scoliosis is far more common in girls than in boys. One girl in 10, but only one boy in 25 has some degree of scoliosis. Most cases have little or no cosmetic or medical significance and never get any worse. Only about 2-3% of cases require medical attention, which most often involves periodic monitoring to determine if progression requires treatment.

Girls are seven times as likely as boys to develop serious spinal curvatures which will require either bracing to halt the progression or surgery to correct it.

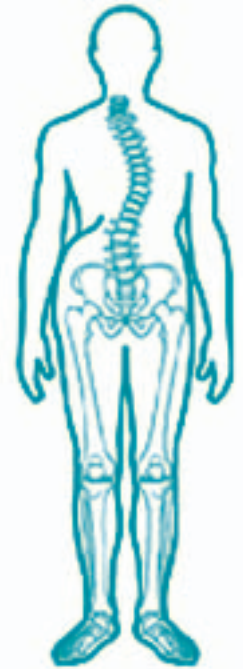
Puberty is the time of greatest risk for the progression of abnormal spinal curves. At that point, while the child's bones are still growing, there is a narrow window of opportunity to take corrective action with bracing that could eliminate the need for surgery. Early detection is critical.

Many states now mandate screening for scoliosis in schools, a practice endorsed by the American Academy of Pediatric Surgeons. Girls should be screened at ages 11 and 13 and boys at age 13 or 14, although the Academy recommends an annual screening for all children 10 through 14. If a parent has scoliosis, it is especially important that his or her children be examined, because the condition runs in families.

Many cases develop in otherwise healthy children, but in 10-20% there is an underlying cause such as a birth defect, a spinal tumor, a neuromuscular disorder such as polio or muscular dystrophy, a connective tissue disorder like Marfan's syndrome, cerebral palsy, spina bifida, or radiation therapy to the spine. Occasionally, a difference in leg length or an abnormality in the hip may result in scoliosis as the child grows.

A spine affected by scoliosis may have one or more curves from side to side. Most often the abnormality is at the level of the chest, but it may also occur in the lower lumbar region or in both parts of the spine. Cosmetic problems such as uneven shoulders or hips, a protruding shoulder blade, or slanted waistline and clothes that don't hang right can be a problem for children just at the age when they are most likely to be self-conscious about their appearance. But the medical problems are far more serious. Severe scoliosis can produce pain, arthritic symptoms, and heart and lung complications resulting from compression of one part of the chest.

The National Scoliosis Foundation (800-673-6922) is a good source of information and support for both children and parents.



Advances in CMT Type 2 Research

By **GARTH NICHOLSON**, Associate Professor, University of Sydney, Sydney, Australia

ATTENTION ALL MAB MEMBERS AND CMT RESEARCHERS:

Remember the deadline for applications for the first annual meeting of the North American Consortium is November 1, 2002. Applications received after this date are ineligible for the registration discount. If you have not received an invitation or have misplaced yours, please call the CMTA office at 1-800-606-2682 and one will be sent out immediately.

(Editor's note: This article appeared in the CMT Australia Newsletter, Volume 41, Winter 2002, and is reprinted with their kind permission. Garth Nicholson is a member of our Medical Advisory Board.)

CMT type 2 is the big unknown area in Charcot-Marie-Tooth research. Although CMT 2 comprises some 30% of all forms of CMT, only one gene mutation has been found causing CMT type 2 in one family. Less than half a dozen chromosomal loci for CMT type 2 have been mapped. This is probably because CMT type 2 is harder to diagnose, occurs with a later onset, and does not occur in large families as often as CMT type 1. CMT type 2 affects the ends of the long nerves, and therefore, like CMT 1, affects the hands and feet first.

CMT type 1 is caused by defects in Schwann cells and the myelin insulation sheath, which is wrapped around nerve fibers. CMT 1 has an element of axonal neuropathy, which appears later as the disease progresses due to an abnormal interaction with the myelin sheath and the nerve fiber, itself. The ends of the long nerves show problems first. This is probably because of difficulties in transporting proteins and nutrients down long nerves.

The first and only CMT 2 gene mutation is a microtubule motor gene, which is a motor involved in carrying proteins down the nerve fiber. If you look inside a nerve axon at high magnification, you see something like a multi-lane freeway, where the lanes are alternating so

the traffic is going both up and down the nerve fiber at the same time.

I recently returned from the International Neuromuscular Disorders meeting in Vancouver, British Columbia, and a Japanese group had reported that they found up to 50 different proteins involved in axonal transport. Some of these are motor proteins. They appear to work like a man with two legs, running along a path while others work like a single leg, carrying protein from the top and running down a rail. It is likely that there are many more genes to be found causing CMT type 2, and many of these recently described axonal transport proteins may be involved.

We have recently carried out a survey of a dozen large families with CMT type 2 to see whether they mapped to the known chromosome loci, already described for type 2. The laboratory work was carried out by two Dutch medical students. They examined the five known chromosomal loci for CMT type 2 and found only one family with one of these known forms of CMT 2. This indicates that type 2 is probably composed of many different molecular varieties, each of which is rare. This will make testing for CMT 2 quite difficult. When multiple genes have to be tested, it will be an expensive exercise. However, when we understand how these genes act, it may be possible to devise treatments to make the defective proteins work better by overcoming some of the axonal transport problems caused by type 2.

DR. GARETH PARRY

(Continued from page 5)

ing that the young man I had seen in New Zealand had CIDP rather than CMT.

Over recent years I have become particularly interested in another inherited neuropathy that is the genetic mirror image of CMT1A, namely hereditary neuropathy with liability to pressure palsies (HNPP). CMT1A results from a reduplication of a segment of chromosome 17 that contains the gene that encodes for a specific myelin protein. HNPP results from a deletion of that segment of chromosome 17. The disorder is usually characterized by recurrent episodes of paralysis or numbness. Each episode resolves, but eventu-

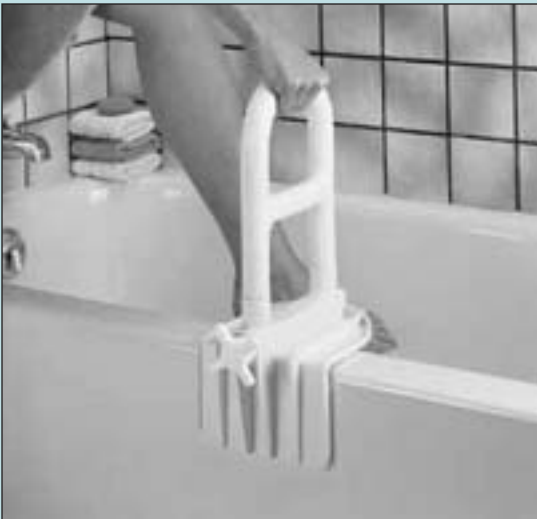
ally there is a progressive neuropathy that can closely resemble CMT. One of the most puzzling aspects of this disorder is the amazing variability that is seen in different patients with an identical genetic defect, and we continue to work with these patients to try to better understand this. The CMTA has now accepted that HNPP should be included as one of the CMT disorders.

Over the more than 30 years of my practice, one of the greatest sources of my professional satisfaction has been my work with people with CMT disorders. It has been a privilege to serve as a member of CMT Medical Advisory Board for more than 10 years, and I look forward to many more years of working with both patients and the Association.

More Gadgets That Make Life Simpler and Safer

Portable Grab Bar

The bi-level grab bar makes it easier for you to get in and out of the tub at home or when traveling. The 15" bar attaches securely to the side of any plastic or fiberglass tub without tools or drilling. It attaches with a large clamp from 3" to 7". It is made of heavy gauge steel with an easy-to-clean nylon coating. Approximately \$85.00 dollars. From Sammons/Preston.



One-Handed Manicure System

A suction cup base holds the platform of the nail clipper/file in place on any smooth flat surface, so you can trim nails with only one hand. Place your fingertip on the front of the platform and press down on the clipper to trim your nails. Excellent for people with weak or shaky hands. About \$25.00 from Sammons/Preston.



Exercise Ball

Squeeze this exercise ball to increase strength and range of motion in fingers, hands, wrists and arms. The microwaveable Therabeads generate moist heat to reduce pain and stiffness. An exercise handbook is included. About \$15.00 from Sammons/Preston.



Long-Handled Brushes and Combs

Body Care long-handled brushes and combs enable persons with limited arm or hand movement to style their own hair. Anti-slip handles are ergonomically designed to lie against the entire palm. They are available in two lengths. From \$13.00 to \$18.00. Sammons/Preston.



Editor's note: The Sammons Preston catalogue, entitled Enrichments, is available by calling 1-800-323-5547 or on the Internet at www.sammonspreston.com

Support Group News

■ California - Berkeley Area

Because the Albany library will not be available during the month of October, the group will meet on November 16th, but the meeting will be tinged with sadness. The group leader, Ruth Levitan, will be stepping down at that time. She reported to the office that her husband's declining health has made the decision a necessity. So far, no one has offered to take over the group, so it may be the final meeting of the support group. As Ruth wrote in her resignation letter, "it has been a rewarding four years for me and I wish I could continue, but 'for everything there is a season and a time for every purpose under heaven' and it's now time for me to move on." Thanks, Ruth, for a job well done!

■ Colorado - Denver Area

The group held its latest meeting on September 9, 2002, with a "Share and Care" meeting. These popular meetings allow members to discuss what works for them and how they deal with the effects of CMT. There was also a discussion of dues and other pertinent issues regarding the operation of the group. Their newsletter highlighted the last issue of The CMTA Report and complimented Steve O'Donnell on his amazing accomplishment in swimming the Chesapeake Bay.

■ Oregon-Willamette Valley/Pacific NW

The August meeting was the annual potluck lunch and was well attended. The group produces and sends to the main office one of the most complete and professional newsletters of any support group. This month's three-page publication includes information about the upcoming meeting as well as general information on adaptive devices, scooters, ramps, and such alternative treatments as Russian Stimulation and massage therapy. A small section discusses the National Peripheral Neuropathy Conference, which was held this summer in Oregon and was organized by Jeannie Porter.

NEW GROUP!!!

■ Western Pennsylvania - Johnstown

A new support group is forming under the leadership of J. D. Griffith. Their first meeting was September 21, 2002, at the Crichton Rehabilitation Center in Johnstown, PA. Featured speakers included Ann Lee Beyer, Chairman and President, and Executive Director, Charles Hagins. If you were unable to attend this first meeting but would like to be informed of future meetings, contact J. D. Griffith at jdgriffith@mail.charter.net.

To demonstrate their support for fundraising events, Charles Hagins, Dick Sharpe, Steve O'Donnell, Ann Beyer, Robert Kleinman, Amar Kamath (Athena Diagnostics), and Phyllis Sanders all attended the Golf Tournament at The Creek.



CMTA Support Groups

Arkansas—Northwest Area

Place: Varies, Call for locations
Meeting: Quarterly
Contact: Libby Bond, 501-795-2240
E-mail: charnicoma57@yahoo.com

California—Los Angeles Area

Place: Various locations
Meeting: Quarterly
Contact: Serena Shaffer,
 818-841-7763
E-mail: serenam71@earthlink.net

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

Place: 300 Sovereign Lane,
 Santa Rosa
Meeting: Quarterly, Saturday, 1 PM
Contact: Freda Brown, 707-573-0181
E-mail: pcmobley@mac.com

Colorado—Denver Area

Place: Glory of God Lutheran Church
 Wheat Ridge
Meeting: Quarterly
Contact: Marilyn Munn Strand,
 303-403-8318
E-mail: mmstrand@aol.com

Kentucky/Southern Indiana/ Southern Ohio

Place: Lexington Public Library,
 Northside Branch
Meeting: Quarterly
Contact: Robert Budde,
 859-255-7471

Massachusetts—Boston Area

Place: Lahey-Hitchcock Clinic,
 Burlington, MA
Meeting: Call for schedule
Contact: David Prince, 978-667-9008
E-mail: baseball@ma.ultranet.com

Michigan—Flint

Place: University of Michigan,
 Health Services
Meeting: Quarterly
Contact: Debbie Newberger/
 Brenda Kehoe,
 810-762-3456

Minnesota—Benson

Place: St. Mark's Lutheran Church
Meeting: Quarterly
Contact: Rosemary Mills, 320-567-2156

Mississippi/Louisiana

Place: Clinton Library, Clinton, MS
Meeting: Quarterly
Contact: Flora Jones,
 601-825-2258
E-mail: flojo4@aol.com

Missouri/Eastern Kansas

Place: Mid-America Rehab Hospital,
 Overland Park, KS
Meeting: First Saturday bimonthly
Contact: Lee Ann Borberg,
 816-229-2614
E-mail: ardi5@aol.com

Missouri—St. Louis Area

Place: Saint Louis University Hospital
Meeting: Quarterly
Contact: Carole Haislip, 314-644-1664
E-mail: c.haislip@att.net

New York—Greater New York

Place: NYU Medical Center/
 Rusk Institute, 400 E. 34th St.
Meeting: 2nd Thursday of each month
Contact: Dr. David Younger,
 212-535-4314,
 Fax 212-535-6392
Website: www.cmtnyc.org

New York—Horseheads

Place: NYSEG Meeting Room, Rt. 17
Meeting: Quarterly
Contact: Angela Piersimoni,
 607-562-8823

New York (Westchester County)/ Connecticut (Fairfield)

Place: Blythedale Hospital
Meeting: 3rd Saturday of each month,
 excluding July & August
Contacts: Diane Kosik, 914-937-2013,
 Beverly Wurzel,
 845-783-2815
E-mail: ladydismiles@aol.com or
 cranomat@frontiernet.net

North Carolina—Archdale/Triad

Place: Archdale Public Library
Meeting: Quarterly
Contact: Ellen (Nora) Burrow,
 336-434-2383

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

Place: Church of the Reconciliation,
 Chapel Hill
Meeting: Quarterly
Contact: Susan Salzberg,
 919-967-3118 (evenings)

Ohio—Greenville

Place: Church of the Brethren
Meeting: Fourth Thursday,
 April–October
Contact: Dot Cain, 937-548-3963
E-mail: Greenville-Ohio-CMT@
 woh.rr.com

Oregon/Pacific NW

Place: Portland, Legacy Good Sam
 Hospital, odd months
 Brooks, Assembly of God
 Church, even months
Meeting: 3rd Saturday of the month
 (except June and Dec.)
Contact: Jeanie Porter, 503-591-9412
 Darlene Weston, 503-245-8444
E-mail: jeanie4211@attbi.com or
 blzerbabe@aol.com

Pennsylvania—Philadelphia Area

Place: University of PA, Founders
 Building, Plaza Room A
Meeting: Bimonthly
Contact: Amanda Young,
 215-222-6513
E-mail: stary1@bellatlantic.net

Pennsylvania—Johnstown Area

Place: Crichton Center for Advanced
 Rehabilitation
Meeting: Bimonthly
Contact: J. D. Griffith, 814-539-2341
E-mail: jdgriffith@mail.charter.net

Ask the Doctor

Dear Doctor,

I have CMT and am 53 years old. I have had 11 operations on my feet, including the last ones which fused my ankles. In the last 8 months, I've started having problems around my knee. When I walk about 10-12 minutes, or stand still for that long, my left leg becomes basically paralyzed. Around the knee region, I lose the use of the leg. Could it be the CMT spreading above the knee? If it is, can you tell me what I can do?

An Orthopaedic surgeon from the MAB replies:

I would strongly suggest that you see your neurologist, preferably the person who diagnosed you with CMT. You should get an updated neurological exam and a manual muscle test of your leg strengths to see whether there is muscle weakness or not.

If this does not produce a satisfactory explanation, then, it is important to re-assess the operations that you have had. It seems that there are a lot and orthopaedists seldom see or recommend fusion of both ankles, unless they were involved in severe accidents, had severe degenerative changes, or had congenital malformations. The position of the ankle (which by definition, after fusion, is stiff) would put additional stress on the knee joint and depending on the attitude of the heel and foot (something that runners frequently refer to) could cause knee problems.

Also, your height, weight, and activity level of work and recreation are also factors to consider.

Dear Doctors,

I know that nerve biopsies can be used to help diagnose CMT, but I thought I read somewhere that muscle biopsies are sometimes also used. Is this true? Any information you would be able to supply would be great.

Several members of the MAB collaborated on this reply:

There is no good evidence that undertaking routine muscle biopsy in CMT is needed. However, special circumstances in which you would consider doing it are:

- If other genetic and acquired disorders are in the differential diagnosis, such as amyloid, sarcoid, mitochondrial disorders, and others.
- If muscle disease is combined with nerve disease, as in some endocrine and vasculitic disorders.

- In some evaluations of infants and very young children where the clinical and electrodiagnostic studies may not give clear distinction between muscle and nerve disorders, especially if general anesthesia needs to be given, anyway, for nerve biopsy.

In general, if an appropriate clinical and electrodiagnostic study indicates that a specific genetic study should be performed, this has priority over nerve biopsy, but both may be needed.

Dear Doctor,

We have been to two orthopaedic surgeons who both suggest surgery for my daughter, who was diagnosed with CMT1B in November 2001. She has had this problem most of her life and has worn AFOs for the past three years. According to her doctors, the type of surgery should depend on the type of CMT. Now, one doctor says it is crucial to have a plantar fascia release done before she stops growing and the cartilage hardens and her bones become deformed. She has a severe arch in both feet and claw toes. Without her AFOs, she walks with great difficulty and turns her ankles. Our concern is the "hurry" to do this operation, the pain, a lack of bettering the situation and the need for later surgeries for her toes. In short, we are looking for the best solution to the problem.

An orthopaedic surgeon from the MAB responds:

There is definitely a "time-frame" in which foot surgery may be more effective in the growing child. The best time is before the foot bones stop growing. After grown stops, the deformity becomes "fixed" and the shape of the bones change, governed by the deforming forces on the foot.

The surgery that may be most effective for a child who has ankles that turn in because of the imbalance is not necessarily a plantar fasciotomy. The strength of the muscles that turn the ankles in and out needs to be known, and, if that is causing the imbalance, tendons may need to be moved to a different location so that it will pull in a more natural direction. Obviously, if the foot has a very high arch, plantar fasciotomy may be helpful, but as a secondary procedure.

Just flattening an arch (which is actually not an easy procedure, no matter which way it is done and which should affect other tissues as little as possible) without knowing what causes it to be "drawn up" or high, does not solve the problem.

The CMTA Report

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The opinions expressed in the newsletter are not necessarily those of the Charcot-Marie-Tooth Association. The material is presented for educational purposes only and is not meant to diagnose or prescribe. While there is no substitute for professional medical care for CMT disorders, these briefs offer current medical opinion that the reader may use to aid and supplement a doctor's treatment.

Letters to the Editor

Dear CMTA,

Please warn others with CMT about the neurotoxic effects of Macrobid. After four years, I have made very little recovery and I have clearly sustained a neurotoxic reaction to this drug. Doctors are still sorting things out, but it caused 1) intracranial hypertension, 2) inflammation of my veins, 3) worsening of my neuropathy, and 4) a blood disorder.

For me, this medication was poison.

Nitrofurantoin, (Furadantin, Macrobid, Macrochantin) is a potentially toxic drug with many side effects. According to a published survey of adverse reactions to the drug, in most cases, peripheral neuropathy begins within the first 45 days of treatment. In 16% of the cases, the symptoms are suspected to develop 2 to 42 days after the medication is discontinued. In rare cases, symptoms may not develop until 160 days after the medication is discontinued.

In an article entitled "Nitrofurantoin-induced Neuropathy and Muscle Weakness: A Case Report with Chronic Use" by Jeffery Fudin and Jenny Allen, the authors comment on the neuropathic pain and muscle weakness encountered by a white male receiving nitrofurantoin for a chronic urinary problem. Short term use of nitrofurantoin is associated with peripheral neuropathy and muscle weakness, but long term use had not previously been studied. They revealed that discontinuing use of the drug resulted in a decrease in pain and a return to normal muscle strength function within four weeks. They advised that the patient never be prescribed nitrofurantoin again.

—G.T. Sebastopol, CA

(Editor's note: Ironically, a second letter regarding Macrobid arrived in the same week's mail.)

Dear CMTA,

I reviewed a response to a question regarding medications and antibiotics. The respondent doesn't mention Nitrofurantoin (Macrobid) which is devastating to CMT people. I would urge your respondent to include this in their response. They may refer to Dr. Thomas Bird of the University of Washington for further information. It has been a common antibiotic prescribed for bladder infections, but it causes sometimes irreversible second neuropathies and progression of the CMT. I am one of the people affected by it.

—M.C. Oregon

Dear CMTA,

Thank you so much for all the information contained in *The CMTA Report* that I receive. The articles are timely and the profiles provide inspiration for me as I battle the challenges of the disease. My interest was especially sparked by the article in the August 2002 issue about the Wayne State CMT Clinic. Could you provide me with a phone number or email address so that I may explore the possibility of visiting the clinic?

—R.K. Denver, PA by email

(Editor's note: The phone number was emailed to him. For others interested in being seen at the clinic, see the article You Can Make a Difference in this issue.)

Dear CMTA,

As a person with CMT, I have suffered with painfully cold feet for years. I finally have the answer to my prayers. Microwave-heated slippers really work! "Cosysoles" heated slippers were designed and developed for a lady with CMT and are available through the website www.cosysoles.ca.

I'm sure that everyone who suffers with cold feet and nerve pain in their toes will find relief from these slippers as I have.

—P.K. by Internet

(Editor's note: The slippers are rather expensive. They list at \$44.95 on the website for adults and slightly less for children.)

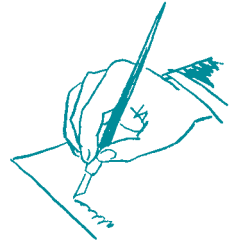
Dear CMTA,

I am sorry that I can no longer afford to send in a donation or membership as I am on disability. I live with my father so that I can afford my prescriptions, to assist him as much as possible, as he, too has CMT. I am the only one that we know of who has CMT and scoliosis. It is bad with a curve of 45° at the top and 70° at the base. I had an operation in 1998, but it didn't work and I still have problems with the rods in my back. The doctor says he has only seen one other spine as bad as mine and he was a boy of 27 years. I'm sure he did better with his surgery because he was younger. I was 51 when they finally operated on me.

I will send some money when I can.

—Z.J. KY

(Editor's note: Members who give more than their \$35 fee to membership allow the CMTA to give this woman a complementary membership so she can continue to receive her newsletter. We thank you and we know she does, as well.)



WRITE TO US!

Pat Dreibelbis, Editor

The CMTA Report

CMTA

2700 Chestnut Pkwy.

Chester, PA 19013

The CMTA reserves the right to edit letters for space.

MEDICAL ALERT:

These drugs are toxic to the peripheral nervous system and can be harmful to the CMT patient.

Adriamycin
Alcohol
Amiodarone
Chloramphenicol
Cisplatin
Dapsone
Diphenylhydantoin (Dilantin)
Disulfiram (Antabuse)
Glutethimide (Doriden)
Gold
Hydralazine (Apresoline)
Isoniazid (INH)
Megadose of vitamin A*
Megadose of vitamin D*
Megadose of vitamin B6* (Pyridoxine)
Metronidazole (Flagyl)
Nitrofurantoin (Furadantin, Macrodantin)
Nitrous oxide (chronic repeated inhalation)
Penicillin (large IV doses only)
Perhexiline (Pexid)
Taxol
Vincristine
Lithium, Misomidazole, and Zoloft can be used with caution.

Before taking any medication, please discuss it fully with your doctor for possible side effects.

*A megadose is defined as ten or more times the recommended daily allowance.



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).
- ... 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).
- ... does not affect life expectancy.
- ... has no effective treatment, although physical therapy, occupational therapy and moderate physical activity are beneficial.
- ... is sometimes surgically treated.
- ... is usually inherited in an autosomal dominant pattern, which means if one parent has CMT, there is a 50% chance of passing it on to each child.
- ... Types 1A, 1B, 1X, HNPP and EGR-2 can now be diagnosed by a blood test.
- ... is the focus of significant genetic research, bringing us closer to answering the CMT enigma.

The CMTA Report

Information on Charcot-Marie-Tooth Disorders from the Charcot-Marie-Tooth Association



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Chester, PA 19013
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www.charcot-marie-tooth.org

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