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DECEMBER 2000

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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CMTA Grant Recipients Present Findings at Boston Neurology Meeting

A satellite meeting was held in conjunction with the American Neurology Association's meetings at the Boston Marriott on October 15, 2000. The one-day session featured a morning devoted to presentations by CMT fellows who discussed the following findings:

- A surgical gene therapy via fetal mouse cerebrospinal fluid
- Assessment of a polymorphic variant in PMP 22 in African-American CMT1A patients
- Quantitation of motor unit fallout electrophysiologically and its relation to weakness in Charcot-Marie-Tooth disorder
- Gene isolation in families with Charcot-Marie-Tooth type 1C.

Following a coffee break, further presentations were made on:

- Successful reversal of demyelination in a transgenic model of CMT1A
- Laryngeal and phrenic nerve involvement in CMT: additional features of CMT2C or CMT1C
- A novel autosomal dominant polyneuropathy with intermediate conduction velocities: a large family with an eight-generation pedigree
- Charcot-Marie-Tooth disease in Poland—three years of molecular genetic analysis
- Right and left hand strength in individuals with HMSN.

These presentations represent the work of our postdoctoral fellows who have done investigations under the mentorship of some of our Medical Advisory Board members.

Following lunch, the topic of the symposium shifted to "The Way Forward," a look by some of



Three of the researchers from Wayne State University in Detroit, MI, Drs. John Kamholz, Emilia Ianakova and Michael Shy, paused following their presentations at the Boston Symposium.

the most well-known CMT researchers at what is going on now in CMT research. Presentations were made by Drs. Michael Shy, Garth Nicholson, John Kamholz, Robert Lovelace, Steven Scherer, Rich Lewis, and others on topics such as "Pathogenic mechanisms of CMT," "CMT disorder as a prototypic heterogenic condition," "Axo-glia interactions in CMT," and "Phenotype/genotype correlation in CMTX."

The day-long event was attended by the members of the CMTA Board of Directors, as well as a large number of the members of the Medical Advisory Board. The day of information sharing ended with a reception that was hosted and sponsored by Athena Diagnostics, the CMTA, and the American Neurological Association.

Board Transitions

By ANN LEE BEYER



VISIT OUR HOMEPAGE

at

www.charcot-marie-tooth.org

The CMTA wishes to thank Jack Walfish, who has completed his term as President of the organization. Jack has given many years of service and countless hours to the CMTA and we deeply appreciate all of his efforts. Since he first was elected to the Board, he has served as Treasurer, Chairman of the By-Laws Committee, and most recently, President. Jack is a loyal and valued supporter and has always been there when the CMTA needed him. It is because of Jack's dedication and commitment that we pulled through the "year of the flood" as painlessly and quickly as we did. Without his efforts, we would not have made it. Thank you, Jack.

Congratulations to Ardith Fetterolf, who succeeds Jack as CMTA President. Ardith, too, gives many hours to CMTA activities each year. Besides serving as Vice President, Ardith has been, and continues to be responsible for many

of our educational outreach programs. This includes making all the arrangements for, and staffing, booths at medical conferences. In addition, she oversees and arranges patient-family conferences, and is in charge of the CMTA Support Group Leaders' Conference next summer in St. Louis, Missouri.

Our new Vice-President, Vincent Bertolino, is a longtime friend and supporter of the CMTA. Vince is a genetic engineer and has an MBA. He joined the Board last year, and recently served as Chairman of the Nominating Committee. At present, Vince is involved in writing a business plan to move us into the 21st Century.

We also welcome new Board members, Meryl Friedman and Robert Kleinman. And we accepted the resignation of J. Rodman Steele, who has been a member of the Board of Directors almost since the organization's inception.

A Message from the New President



As I assume the office of President of the CMTA, my goals remain unchanged. I want to help those with CMT in any way that I can. In the years that I have been on the Board of Directors of the CMTA, I have organized family conferences, manned CMTA exhibits at medical conventions, worked to organize support groups and been the leader of one, and served in two other Board officer positions.

I have CMT, being a member of the fourth generation known to have inherited this genetic disorder. There are 8 living members of my family, including aunts and cousins who have CMT. I became very aware of the large numbers of people affected by CMT when I attended my first family conference in Chicago many years ago. Since then, I have met many others at family con-

ferences and have truly seen "the many faces of CMT."

It is difficult to narrow my key interests in the future work of the CMTA, but my primary concerns are the ongoing genetic research and the more practical clinical work, both of which are very important to all of us. These past 17 years since the CMTA was founded have seen great progress in the education of both medical professionals and patients. I want this to continue.

After my retirement 12 years ago from the Department of the Defense, I have been free to spend my time as a volunteer. How better could I spend it than to continue the work of the CMTA? As President, I will continue to help the organization move forward with our Mission with the full support of the Board of Directors and each of you, our members.

Ardith Fetterolf
President



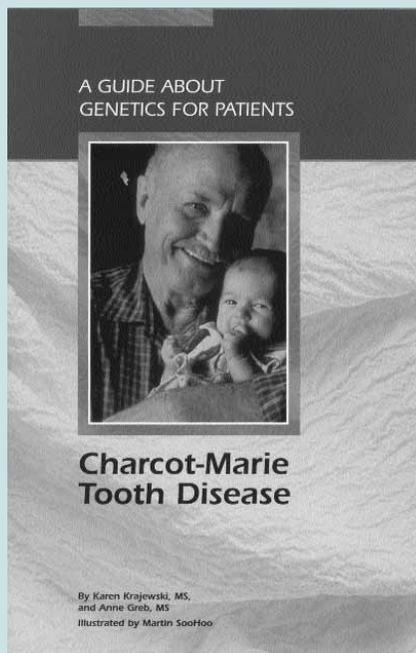
CMTA MEMBERSHIP/ORDER FORM

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.



GENETICS BOOKLET Now Available

The long-awaited booklet on the genetics of CMT is available from the CMTA for the price of \$4 for current members and \$5 for non-members. All proceeds from the sale of this booklet will go to fund research to help in the ultimate cure of CMT.

Not only does the booklet explain, in detail, about CMT1A, 1B, CMTX, and HNPP, but it also explains autosomal recessive inheritance patterns. Because the type 2 variations of CMT have not yet been linked to the causing genes or the specific mutations, those forms are not discussed in detail.

Some commonly asked questions, such as "If someone in my family does not have symptoms of CMT, can he or she have a child with CMT?" or "Why am I the only one in my family with CMT?" are covered in the closing pages of the booklet.

In a compact and easily read format, this little booklet, "A Guide About Genetics for Patients: Charcot-Marie-Tooth Disease" is a must-have for every patient library.

	QTY	COST	TOTAL
Charcot-Marie-Tooth Disorders: A Handbook for Primary Care Physicians		active members \$15 inactive members \$20	
Membership Dues		\$35	
SPECIAL OFFER FOR ACTIVE MEMBERS ONLY: CMT Facts Series (I-IV)		active members \$16	
CMT Facts I <input type="checkbox"/> English <input type="checkbox"/> Spanish		active members \$3 inactive members \$5	
CMT Facts II <input type="checkbox"/> English <input type="checkbox"/> Spanish		active members \$5 inactive members \$7	
CMT Facts III		active members \$5 inactive members \$7	
CMT Facts IV		active members \$8 inactive members \$10	
A Guide About Genetics for the CMT Patient NEW! <i>No shipping and handling on this item only.</i>		active members \$4 inactive members \$5	
VCR Double Tape: duPont Conference		active members \$25 inactive members \$40	
CMT Informational Brochure <input type="checkbox"/> English <input type="checkbox"/> Spanish		FREE	Send Self- Addressed Stamped Envelope
Physician Referral List: States: _____		FREE	
Letter to Medical Professional with Drug List		FREE	
Contribution to CMT Research <i>10% will be applied to administrative expenses.</i>			
Shipping & Handling <i>Orders under \$10 add \$1.50, orders over \$10 add \$3.00</i>			
TOTAL			

Check payable to the CMTA (US Residents only).
Foreign residents, please use a credit card or International Money Order.

VISA MasterCard American Express

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Signature _____

Mail to the CMTA, 2700 Chestnut Parkway, Chester, PA 19013

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.

Orthotic Management and Treatment of CMT-Affected Limbs

By **BRUCE GOODMAN, Certified Orthotist, Progressive Orthotics, Selden, NY**
 Illustrations by Reuven Gayle

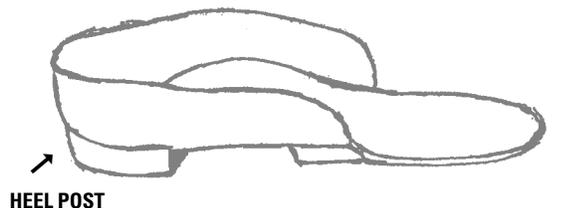
Orthotic management of patients who suffer from Charcot-Marie-Tooth disorders needs to address the individual's specific neuromuscular weaknesses and the problems that are common throughout this population. This understanding enables the orthotist to recommend the simplest orthosis that can be effective. Proper foresight in understanding how the disease may progress is very important in determining when prescription considerations need to be modified.

Early orthotic intervention decreases some of the forces that are placed onto the ankle and foot due to cavus deformity (hollow foot, an exaggeration of the normal arch). The increased likelihood of routine ankle sprains can be minimized with a simple orthotic or UCBL insert with a lateral heel and forefoot post (see figure below). A UCBL insert is an orthopedic appliance that is a compromise between an arch support and an orthosis. This device fits into one's shoe, but the uppermost trimline is concealed by the top of the shoe. This allows the UCBL greater control of the calcaneus (heel bone) than a simple arch support. These devices help to shift one's body weight more directly over the ankle joint. This more stable position will help to reduce the repeated trauma that often leads to arthritic changes later in life.

The ankle, knee, and hip are designed to work with one another to shorten the limb to allow floor clearance during the swing phase of walking. When the dorsiflexors, which turn the foot or the toes upward, are no longer functioning correctly due to peroneal nerve weakness, a steppage gait occurs. This gait pattern helps the person to avoid tripping; however, other biomechanical ramifications need to be considered. The subtalar joint allows side-to-side motion in the feet and ankles. This joint is positioned in such a way that when the foot points downward, it automatically inverts (turns inward). This position places more weight on the outer border of the foot. This is the same biomechanical problem presented by the cavus deformity discussed earlier. This compounds the problem by placing the foot in an even more vulnerable position, since many patients already have instability due to their cavus foot deformity.

The orthotic appliance of choice to help control this problem is a custom-made plastic ankle-foot-orthosis (AFO), which allows free plantar flexion, with a dorsiflexion assist. This orthosis should include a lateral flange and soft interface as well as a dorsum strap. A heel and forefoot post might be necessary in some cases. This orthosis is fastened with a Velcro closure.

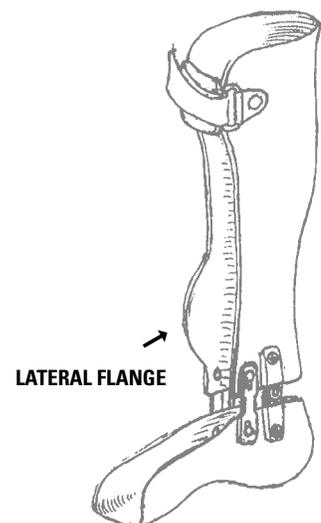
UCBL INSERT



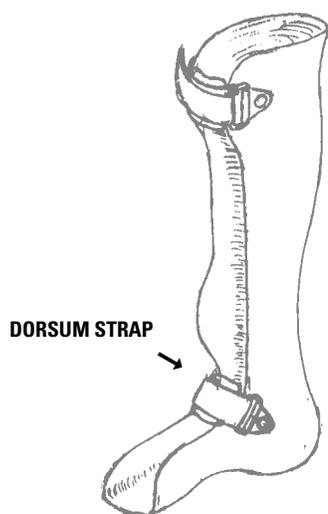
ORTHOTIC



ANKLE-FOOT-ORTHOSIS (HINGED)



SOLID-ANKLE-FOOT-ORTHOSES



The key element to this bracing recommendation is that this brace contains a flexible ankle joint, allowing all other muscles to fire normally while assisting only the weakened dorsi-flexors. The lateral flange and heel and forefoot post help to redirect the forces more toward the center of the ankle joint, increasing stability and reducing the forces adding to the deterioration of the ankle joint.

As arthritic changes advance within the joint, the ankle often becomes rigid, with little or no motion taking place. Sometimes, this could also describe a joint that has been surgically fused as well. The orthosis indicated at this time is a custom-made plastic solid-ankle-foot orthosis, which has no hinge. This brace can also contain a lateral flange, soft interface, dorsum strap, and heel and forefoot posting, if necessary. This brace is also applied with a Velcro closure.

This design immobilizes the foot and ankle, transferring some of the forces from the ankle to the brace and helping control the weight line to a more stable position (see figure above).

It is important that a thorough orthotic evaluation take place prior to design considerations. This should include manual range of motion as well as manual muscle testing of the involved limb. The contralateral or opposite limb's health and strength need to be considered as well. In addition, the patient should be observed and studied while walking. This gait analysis is of utmost importance in determining the biomechanics of the individual patient. As important as the prescription is, the fitting and follow-up care are also essential for successful orthotic management.

The best possible results are obtained when there is effective communication between the patient, the doctor, the physical therapist, and the certified orthotist.

Pedorthic Management of the CMT Patient

By **STEVEN RUEDA, Certified Pedorthist,**
Turnpike Comfort Footwear, Flushing, NY

For those of you not familiar with the practice of pedorthics, it is the design, manufacture, fit, and/or modification of shoes and foot orthotics to alleviate foot problems caused by disease, overuse, or injury. Board-certified pedorthists are trained in foot anatomy, biomechanics, and the construction of shoes and foot orthotic devices. Certified pedorthists are trained to select proper footwear for specific foot conditions and ensure that prescriptions perform correctly for the patient, according to the doctor's intentions.



Patients with Charcot-Marie-Tooth disorder must understand that wearing quality footwear that is biomechanically correct is of significant value to the individual's foot health. Properly fitted shoes improve orthotic and bracing treatment, and poorly chosen shoes compromise treatment.

Applying footwear to CMT-afflicted feet requires the basic understanding that the shape of the shoe must conform to the shape of the foot. The foot and shoe should work together to reduce irritation to the foot and improve shoe function.

The challenge to the pedorthist in fitting patients with CMT is accommodating the high arch, claw or hammer toes, and inversion (rolling out) of the foot. In early onset of the disease, this can be done simply with extra-depth shoes or sneakers and some wedging of the heel and sole of the shoes to stabilize the foot from inverting out. As the disease progresses, so does the need for more support. This can be accomplished with an orthotic device (arch support), which will help distribute weight and add more stability. In addition, high-top shoes and/or buttresses (supporting walls added to the outside of the shoe) may be needed. Custom shoes may be necessary in cases of more serious deformity. Ultimately, lower-extremity bracing may be needed, which is performed by the certified orthotist.

Some of the footwear products that CMT patients might find helpful are extra-depth shoes by P.W. Minor, which are available in EEEE and EEEEE widths, and which offer long counters for more support and stability and are perfect to accommodate orthotics and braces. They also have introduced a line of shoes called Xsensibles, which have a revolutionary five-layer upper material that stretches 70% horizontally and 30% vertically—perfect for hammer toes.

New Balance sneakers come in EEEE widths, have high toe boxes, and have removable inserts to accommodate orthotics and braces.

There are many other products that can be functional for the CMT patient. However, the most important thing to remember is to find a qualified certified pedorthist to assist you in your footwear selection and prescription needs. To find a pedorthist in your area, call the Pedorthic Footwear Association at 1-800-673-8447 or see their web site at www.pedorthics.org

CONTINUING OUR EDUCATIONAL PROGRAMS

University of Pennsylvania Hosts CMT Patient Conference

By PAT DREIBELBIS

On Saturday, October 7, 2000, over 100 members and friends of the CMTA attended an all-day conference on Charcot-Marie-Tooth disorders at the BRB Auditorium on the campus of the University of Pennsylvania School of Medicine. The conference was organized by the new CMTA President Ardith Fetterolf and Dr. Steven Scherer, neurologist, from the University of Pennsylvania.

The morning began with a panel discussion on the current status of CMT research by some of the pre-eminent researchers in the United States: Drs. Lisa Baumbach-Reardon of the University of Miami, Phillip Chance from the University of Washington, Jeffrey Vance of Duke University, and Steven Scherer from the University of Pennsylvania. They each discussed one of the variants of CMT, including the African-American variant of type 1A, type 2, HNPP (hereditary neuropathy with liability to pressure palsies), and X-linked CMT.

Dr. Jeffrey Vance gave an excellent talk on type 2, about which less is generally known and written. He mentioned that type 2 patients generally have fewer sensory symptoms and

Three of the morning presenters at the Philadelphia patient/family conference were Dr. Jeffrey Vance, Duke University, Dr. Lisa Baumbach-Reardon, University of Miami, and Dr. Phillip Chance, University of Washington.



Attendees of the Philadelphia conference listen to a presentation by Dr. Carol Oatis on physical therapy and exercise for CMT patients.

less upper extremity weakness. Approximately one-third of all cases of CMT are type 2. When doing a nerve conduction velocity, the dividing line between types 1 and 2 is usually a score of 38 m. Below that is type 1 and above is type 2. He informed the audience that CMT2A is located on chromosome 1; 2B is located on chromosome 3 (this type is extremely rare); 2C causes vocal cord weakness and respiratory failure, but the location is not known yet; and 2D is located on chromosome 7.

Dr. Scherer discussed CMTX, which is the second most common form of CMT. It is like CMT1, except that males are affected earlier and more uniformly than females. It is caused by the connexin32 gene and patients with CMTX typically have nerve conduction velocities in the range of 25-35 m/second. Type X shows demyelinated and remyelinated fibers, onion bulb formations, and more axonal loss than is typical in 1A or 1B.

The questions from the audience that followed these presentations were challenging and reflected the increased knowledge that patients have about their own disorder.

After a short break, Ann Greb, MS, one of the authors of the new genetics booklet, spoke about genetic counseling and the value it can provide to families dealing with CMT. She explained that a genetic counselor will review



a family's medical history and listen to their concerns about conditions affecting the family. Genetic conditions are not rare. They afflict about 10% of adults and 30% of children, in the hospital. A counselor does not make decisions for a family, but rather encourages them to make decisions that reflect their own personal and cultural beliefs with regard to disease and inheritance. Each person at the conference received a copy of the new booklet to take home.

Other presentations included a brief overview of genetic testing that is available from Athena Diagnostics, a comprehensive discussion of physical therapy options in dealing with CMT by Dr. Carol Oatis, the author of one of the chapters in the physician's handbook on CMT, a discussion of the many

options for handling the pain associated with CMT, and a detailed presentation on foot and ankle procedures by Dr. Jason Miller, DPM. He stressed that the treatment that is best at one stage in the disease process is not necessarily the best at a later stage or for another person. Each person should be individually evaluated, and the least invasive but most useful treatment should be suggested first.

The auditorium was a lovely new facility with comfortable chairs; the food that was served was unusually good; the presenters were generous with their time and expertise. The only complaint that was heard the entire day was that the facility had been difficult to find. Many people emailed or called the office following the conference to express their pleasure at having attended.

Children's Books Feature Heroine with CMT

Pamela J. Walls, a long-time member of the Charcot-Marie-Tooth Association and a freelance writer, has been trying for years to get an article on CMT published. Now, she has found a way to spread the news about CMT through the publication of the first two of a series of eight novels about a young heroine who has CMT. Her main character is a 13-year old named Abby, whose best friend is a 14-year old named Luke Quiggley.

Together, these two get themselves into and out of some curious adventures in and around Hawaii. The series is called "South Sea Adventures" and features the first novel, *Abby: Lost at Sea*, and the second, *Abby: Quest for Treasure*. In the first novel, Pam includes information about her having CMT in the "about the author" section and also refers readers to the Association for further information.

The cover artist is Jean Paul Tibbles, who is well-known for his work on the



American Girl covers. He brings Abby to life on the cover and makes her easy for the reader to imagine and enjoy. Pam describes her heroine as "a spunky heroine who overcomes her physical limitations with intelligence, courage and compassion. While facing all kinds of danger—pirates, sharks, stormy seas and villains—Abby always comes out on top!"

The books have religious and moral overtones, and in virtually every danger Abby turns to her faith in

God to help her with decisions and victories. The publisher, Tyndale House, is known for its "Left Behind Series," which has sold more than 12 million copies. They have recently expanded into children's fiction and Pam's books are part of their initial offering in that arena.

Pam closes her comments with the hope that "they [the books with Abby as heroine] encourage our many CMT kids across the nation."

1999-2000: A Very Good Year for the CMTA

In spite of the notorious flood of 1999, this has been a good year for the CMTA. During the past year, we have been a presence, staffing booths, at the American Neurological Association, the American Academy of Neurology, The American Society of Human Genetics, The American Podiatric Association, the Physical Therapy Association, and the Podorthotics Conference. Every meeting at which we have a booth and distribute literature is an opportunity for more professionals to become aware of our organization and the educational materials that we offer.

In addition, we arranged two patient/family conferences on opposite sides of the country: one in Salt Lake City, Utah, at the University of Utah and a second one, more recently, at the University of Pennsylvania in Philadelphia, PA. Each of these conferences attracted families anxious to learn more about living with CMT and what promise research holds for the future.

We were fortunate to have two golf fundraisers, which took place in New York, and which raised over \$37,000 together for the research fund. We also received the largest single gift ever to the organization from the estate of James Thomas Moore. Because of these special circumstances, we find ourselves in a good financial position to make research grants.

We have recently published a new genetics booklet, which should help patients and family members understand a rather complicated subject.

The Board of the organization represented the membership at a satellite meeting on recessive forms of CMT, found commonly in the Mediter-

The meeting on recessive CMT, in Italy, was organized by Dr. Lefkos Middleton and attended by a group of European and American researchers, who assembled on the steps of the hotel.



Dr. Georges Serratrice attended the conference on recessive CMT forms and is best known for having arranged and hosted the first International Conference on CMT Disorders in 1976 in Marseille, France.

ranean basin area. The meeting was held in Italy, where we learned more about the biology of CMT and made contacts and formed alliances with European CMT clinicians and researchers. We also attended the satellite meetings in Boston, which are discussed on page 1 of this newsletter.

Work continues in the discovery of new genes responsible for forms of CMT and we funded three postdoctoral fellows whose work in genetic research was reported in Boston. Two doctors from Italy, Paolo Vinci and Sandra Perilli, who have designed a boot to help people with CMT walk more comfortably, presented their findings to a group of doctors and rehabilitation specialists at Columbia.

Just recently, the Chairman, President, and Vice-President attended meetings on nonprofit board development in Washington, DC, to help the Board better serve the membership.

As successful as we feel this past year has been, we have ambitious goals to make the next year even better. We hope to fund more research grants, establish a CMT database at the University of Indiana, develop CMT Centers of Excellence, sponsor four patient/family conferences, publish a newsletter exclusively devoted to children and CMT, work on a pamphlet about children, CMT, and the school system, and have a national support group leaders' conference.



GIFTS WERE MADE TO THE CMTA...

IN HONOR OF:

Stephanie DiCara

Mrs. Arden Kolb-DeBolt
The Students at Our Lady of the
Wayside School

Jim and Barbara Leisek's 50th Anniversary

Alfred and Carrie Jean Adler
Lee and Jayna Bell
Barbara and Jack Conrad
Jane and John Creveling
Ben and Jean DeCook
Lew and Elaine Elford
Carol and Bill Gilleland
Mrs. Eleanor Mossman
David and Rebecca Orlando
William and Joyce Schrack
Bill Turner and Barb Jones
Carol Van Den Branden

School Donates to CMTA

In the fall 2000 issue of *The CMTA Report*, a letter to the editor featured a tribute to Stephanie DiCara written by her cousin, Jennifer Naye. Since then, Jennifer, who is the Commissioner of Finance at her school, Our Lady of the Wayside, in Arlington Heights, Illinois, has convinced her school to recognize the CMTA as the recipient of one of their monthly charitable gifts. Her efforts and those of her classmates resulted in a gift of \$811.20 being sent to the CMTA.



Jennifer turned her concern for her cousin into a gift for the CMTA.

IN MEMORY OF:

Meryl Baughers

Mr. and Mrs. James Cramer

Samuel C. Cantor

Mr. and Mrs. Eddy Cantor

Jane L. Chase

Marcia K. Potocny

John Curran

West Chester County, NY,
Support Group

Dorothy Eade

Ronald and Linda Simpson

Mike Goldman

Marcia Siegel

Margaret James

Nancy and Gerald Hickey

Jack Kearney

Nancy Gothier

Jacob, Yetta, and Maurice Silver

Henry Silver

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: _____

Creatine: Muscle Builder or Health Breaker

(These comments are from the January 2000 Harvard Men's Health Watch.)

Creatine is an amino acid, one of the 20 that serve as building blocks for all the body's proteins. Creatine is present in many human tissues, including the brain, eyes, heart, and testicles. But for athletes, the most important role is in muscle, where 95 percent of the body's creatine is stored.

To do their work, muscles need energy. That energy depends on adenosine triphosphate (ATP), but muscles store only enough ATP to power 10 seconds of maximal exercise—enough time, perhaps, to slug a homer, but not enough to do much else. To continue exercising, muscles must generate ATP as they go. Creatine has an essential role in producing ATP. First, creatine is transformed into phosphocreatine. Next, it gives up the phosphate, which becomes the P in ATP, and returns creatine to its natural state so it can generate more ATP as needed.

GOOD NEWS...TAX RELIEF

On May 8, 2000, the Internal Revenue Service (IRS) issued a ruling that will allow parents to deduct some of the costs associated with attending medical meetings related to their child's health condition. Congressman George Miller (D-CA), who called for the IRS decision, noted that physicians, vendors, salespeople, and others can deduct the costs of attending medical meetings, but until now, parents could not. The new ruling allows parents to deduct, "amounts paid by an individual for expenses of admission and transportation to a medical conference relating to the chronic disease of the individual's dependent." For further information, see: Internal Revenue Bulletin 2000-19 (May 8, 2000), Rev. Rul. 2000-24, pp.963-964.



Creatine is essential to health, and the body produces 1 to 2 grams daily, primarily in the liver, kidneys, and pancreas. And, if that's not enough, the average man gets another 1 to 2 grams each day from his diet, mostly from meat, fish, and milk. Like other amino acids, excessive creatine is secreted in the urine.

Unlike most sports supplements, creatine has been tested in exercise labs, and it does seem to work. But, before you head to the store or log on to the Internet to purchase it, consider the very special circumstances of these experiments and the very limited benefits of creatine. Most of the studies tested very large doses of creatine, typically in the range of 20 grams a day—about the amount in 10 pounds of steak. All of the studies were brief, lasting just one to eight weeks, and most of the subjects were highly trained young men, usually elite athletes. Under these conditions, creatine supplements appear to increase body weight—but much of the gain is water, not muscle mass. Still, creatine does seem to increase muscle performance, but only for very brief, high-intensity, repetitive tasks such as weightlifting, and only to a modest degree, perhaps 4 percent. Away from the lab, field trials of creatine have found little or no effect on actual athletic performance, and aerobic exercise does not benefit at all.

Creatine may produce marginal gains for certain highly competitive power athletes, but its greatest promise may lie at the opposite end of the performance spectrum, in patients with serious neuromuscular diseases that sap muscle strength. In 1999, scientists in Canada reported the results of a study of 102 patients with such diseases. In two trials, the patients took 10 grams of creatine daily for five days, followed by 5 grams a day for another five to seven days. Strength testing was performed before and after the supplements: on average, treatment appeared to improve muscle strength, in some cases by more than 10 percent. No side effects were observed, but the study was very brief. Although these results are encouraging, much more research is needed before creatine can be recommended for patients with neuromuscular disease.

Ask the Doctor

Dear Doctor,

I am 46 years old and have Charcot-Marie-Tooth disease (CMT). For about five years, I have been wearing AFOs all day. My ability to stand still for any length of time has gotten significantly worse. What I mean by not being able to "stand still" is that I lose my balance. I have no problem whatsoever with getting stiff or with my legs being restless. My thinking is that wearing AFOs all the time might somehow be contributing to additional atrophy of my calf and foot muscles, which I presume is what causes balance problems. Does wearing AFOs have any effect on how rapidly muscle tone is lost? Also, will exercise on a "health rider" (something like a stationary bike) be helpful or harmful? Finally, you should know that I've gained a fair amount of weight after I quit smoking.

—(From the Internet)

An MDA Clinic Director replies:

Loss of balance while standing isn't uncommon with CMT because of loss of proprioception in the joints of the lower extremities starting with the ankles. Wearing AFOs may provide some stability. Another possibility is just wearing elastic or neoprene anklets. The use of braces does contribute to additional atrophy in the calf and foot muscles. Muscular activity is required for protein synthesis and what you don't use, you lose. There is always a "trade off" between atrophy and stability. The question is how vital is the need for stability and will the AFOs provide this? This can only be determined by an appropriate examination and clinical trial. In any event, you should have the orthoses evaluated. They may require refitting or modification.

Use of a "Health Rider" wouldn't be harmful. High-repetition, low-load exercise, as long as it's not continued to the point of exhaustion, should help maintain muscle tone. Such exercise may also help with your weight problem. However, you should be following a well-balanced, but low-calorie diet because of your relatively sedentary state. A consultation with a dietician would be helpful.

Editor's Note: If you have a medical question about CMT, you can mail it to "Ask the Doctor" at the CMTA office (2700 Chestnut Parkway, Chester, PA 19013) or go on-line to www.mdausa.org and choose "Ask the Experts".

Year-End Gift Ideas

When considering year-end tax planning, you should make use of the income tax charitable deduction. A year-end gift can significantly reduce your income taxes while providing meaningful support for the Charcot-Marie-Tooth Association.



Regardless of your income, if you itemize, you can almost always lower your income taxes through charitable giving. The amount of the income tax savings depends on your tax bracket. If you are in the 31% income tax bracket in 2000 and you itemize, a \$1,000 gift to the CMTA by December 31 will save you \$310 in your 2000 taxes.

You might consider making larger charitable gifts in years in which you have the most income or are in the top federal tax income brackets. You should always check with your tax advisor or financial planner. Giving, is, however, much more than tax brackets and charitable deductions. Your gift, no matter its size, makes an important difference in what we are able to accomplish.

You can benefit from the charitable tax deduction by writing a check by December 31. There is no easier way to earn a deduction. Just make sure your envelope is postmarked by December 31. If it is, your gift will qualify even if we do not receive the gift until the first week of 2001. If your employer has a matching gift program, enclose the form with your check and your gift is immediately increased. Gifts of cash are fully deductible, up to a maximum of 50% of your adjusted gross income.

If you own stock, it is almost always more tax-wise to contribute stock than cash. A gift of appreciated stock generally offers a two-fold tax saving. First, you avoid paying capital gains on the increase in value of the stock. Second, you receive an income tax charitable deduction for the full fair market value of the stock at the time of the gift. If you purchased stock some years ago for \$1,000 and it is now worth \$10,000, an outright gift of that stock would result in a charitable deduction of \$10,000 and the avoidance of a capital gains tax on the \$9,000 in appreciation.

You must have owned the stock for a "long-term," which usually means more than one year, in order to qualify for the tax advantage. Again, the gift of stock must be postmarked before December 31. Gifts of appreciated stock are fully deductible up to a maximum of 30% of your adjusted gross income.

Mentioning the CMTA in your will is another way to continue the work of the association. To make a gift to the organization in any of these ways, you should consult your tax advisor to learn how this information relates to your individual circumstances.

We appreciate your support at year-end and throughout the year.

My Experience with CMT

By MYRTLE BROWNE (reprinted from the newsletter of the North Coast Counties, CA support group)

I was diagnosed with CMT in 1978 at the age of 65. As a child and young adult, I didn't seem to have any problems. I walked three miles to and from school. I also did a lot of roller skating, swimming, horseback riding, and other sports with no trouble at all.

In 1977, I started to notice that my ankles were getting weak and I was not stable on my feet. I complained about this to my doctor. He could not find anything wrong, but did send me for a circulation test. An orthopaedic doctor said my left leg was shorter than the right one, so he

suggested wearing a 1/4" lift in my left shoe. Of course, this did no good.

When I saw the head of orthopaedics, he took one look at my feet and high arches and said he was sure I had CMT, but wanted me to see the physical medicine department and a neurologist. I had tests that showed low nerve conduction. Both doctors I saw confirmed CMT. This all took about a year.

I later started having trouble with my balance and had to lean against the wall for support and to keep from falling. In 1981, I tried a pair of ready-made braces. They seemed to help to stabilize my ankles.

In 1986, I was sent to Vallejo for a gait analysis, which didn't turn out too well. They suggested a tendon transfer on my left foot, which I had. It has helped a great deal to keep my toes up so that I don't trip so easily. I didn't have the right foot done because of poor circulation in that leg.

In 1991, I started wearing custom-made AFOs and I'm still wearing them.

I never knew of anyone in my family having CMT, but when I think back, I suspect that my mother might have had a light case of it. She had one very crooked foot and had balance problems in her later years. One of her sisters also had a great deal of foot trouble, but in those days, there was very little known about this disease and no one was ever tested.

So far, my daughter and grandchildren have not shown any signs of it. However, one of my great nephews has been diagnosed as having it. This would indicate that it does come down in my mother's family.

I have had many falls, resulting in two fractured wrists, a fractured foot, and bruised ribs. The last fall injured my back and landed me in a rehab hospital for two weeks. I think my biggest problem is that I try to turn around and move too quickly.

I am able to move around in my home without the use of a walker or cane as long as I am wearing my shoes and braces. In the evening, I usually put on my slippers and then I use the walker. If I go out with my husband, I can get by with just a cane. If I go some place by myself, I always have a walker in my car.

I am now 87 years of age and although my hands are getting pretty stiff, I feel fortunate that I have done as well as I have this long.

CMTA Support Group News

■ Kentucky/Southern Indiana/Southern Ohio

The group reports that the last meeting in November was very successful, with 8 regular members, 3 new members, and a speaker and two assistants. Greg Pits, an occupational therapist, brought a large selection of mechanical devices that are designed to help with simple daily chores such as buttoning shirts, picking up objects, handling change, etc. He will be invited back for another session in 2001. Dr. William Weitzel will be presenting at the next meeting, but the date has not yet been chosen.

■ Massachusetts - Boston Area

David Prince reports that the group had been meeting the first Tuesday of every other month, but has found that, after three years, the attendance has dropped off sharply. The group has over 60 members, but they are not interested in attending unless there is a scheduled speaker. The group, will, therefore, meet next on April 3, 2001 at the Lahey Clinic in Burlington, MA, with plans to meet again on October 2, 2001.

CMTA Support Groups

Alabama/Greater Tennessee Valley

Place: ECM Hospital, Florence, AL
Meeting: Quarterly
Contact: William Porter, 205-767-4181

Arkansas—Northwest Area

Place: Harvey and Bernice Jones Center for Families, Springdale
Meeting: 3rd Saturday of each month
Contact: Libby Bond, 501-795-2318
E-mail: charmicoma@netzero.net

California—Berkeley Area

Place: Albany Library, Albany, CA
Meeting: Quarterly
Contact: Ruth Levitan, 510-524-3506
E-mail: rulev@pacbell.net

California—Los Angeles Area

Place: Various locations
Meeting: Quarterly
Contact: Serena Shaffer, 818-841-7763
E-mail: SerenaM71@aol.com

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@home.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

Florida—Boca Raton to Melbourne

Place: Upledger Institute, Palm Beach Gardens
Meeting: Quarterly
Contact: Cynthia Gracey, 561-243-0000

Florida—Miami/Ft. Lauderdale

Place: North Broward Medical Center, Pompano Beach, FL
Contact: Al Kent, 954-742-5200 (daytime) or 954-472-3313 (evenings)
E-mail: marbearwld@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—Detroit Area

Place: Beaumont Hospital
Meeting: Three times each year
Contact: Suzanne Tarpinian, 313-883-1123

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: Betty Aultman, 601-825-5626
 Julia Provost, 601-825-6482

Missouri/Eastern Kansas

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

Missouri—St. Louis Area

Place: St. Louis University Medical Health Center
Meeting: Quarterly
Contact: Carole Haislip, 314-644-1664

New York—New York City

Place: NYU Medical Center/Rusk Institute
Meeting: Monthly
Contact: Dr. David Younger, 212-535-4314, Fax 212-535-6392

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: Monthly, Saturday
Contact: Kay Flynn, 914-793-4710
E-mail: alma622@worldnet.att.net

North Carolina—Archdale/Triad

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

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

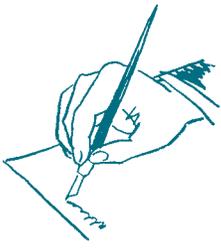
Place: Church of the Reconciliation, Chapel Hill
Meeting: Quarterly
Contact: Susan Salzberg, 919-967-3118 (evenings)

Ohio—Greenville

Place: Church of the Brethren
Meeting: Fourth Thursday, April–October
Contact: Dot Cain, 937-548-3963

Oregon—Willamette Valley/Pacific NW

Place: Alternates between Brooks Assembly of God Church and Legacy Good Samaritan Hospital, Portland
Meeting: Third Saturday of the month
Contact: Jeanie Porter, 503-591-9412
 Darlene Weston, 503-245-8444



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.*

Letters to the Editor:

Dear CMTA,

I have recently been diagnosed with CMT. Of course, it is a frightening reality. I can't believe that I have a "disease." The word disease is such a scary word. My wife and I have been taken aback.

I've always had foot and ankle problems, but I thought the serious problems would come later on in life. I'm only 26 years old.

At first I was angry because the doctor was so blunt. He didn't have a very good bedside manner. Secondly, I got sad thinking about all the things I will eventually not be able to do. I have now accepted the fact that I have CMT.

The biggest support is having a loving and supporting wife. My wife has gone out of her way to help me and stand beside me. We are truly a team.

I would like to learn more about my CMT through your organization. I want to join, as well.

—C.R. Jackson, TN

Dear CMTA,

I am 63 years old and was diagnosed at age 20 with CMT. My doctor told it was incurable; it could only go to my knees and elbows and could put me in a wheelchair. Neither of my parents exhibited any symptoms and I am an only child. My wife and I have been happily married for 46 years. We have three children (44, 42, and 38 years old) and 4 grandchildren (22, 20, 15, and 12 years old). None of them exhibit any symptoms of CMT to date.

At the age of 20, I was an automotive mechanic and experienced a bout of bone-aching flu. After the usual week or so of aches, fever, and feeling bad, I began to feel better for a day or so and then my legs, below the knees, began to ache terribly. I told my wife I must be having a relapse of the flu. After a few days, the pain subsided and I went back to work. About 4 to 5 months later, one of my fellow mechanics told me he noticed that I was walking "differently." I guess I had not noticed, but after that I did notice that I could not stand on my toes or wiggle them. This was when my father and I went to a specialist and I was diagnosed. I've always felt that the flu episode set off the CMT.

I progressed to problems with my hands. I guess that being a mechanic then and for the next ten years was good for me, as I did a lot of

getting up and down under cars and gripping wrenches. I eventually progressed to desk jobs, but all of them required a certain amount of walking, traveling, communicating, etc.

After I was diagnosed, I never looked back and life has been great to date. I retired as Manager of Equipment Engineering for the Dallas Area Rapid Transit Authority in 1995 after 38 years there. I have probably had only 10 to 15 people ever ask me what was wrong with my knee. I would explain. My level of CMT has never hampered me in my work and no one I have ever worked with has ever had a problem with me.

As my HMO company changes, my doctors change and usually at my first meeting, I inform them about CMT. Some seem to know what it is; some don't. I was glad to receive your list of problem drugs for CMT patients, as I have never had any previous information. I am going to give it to each doctor whenever I have to change to a new one. I am blessed with no other known physical problems other than a shellfish allergy and a mountain cedar dust allergy.

As I walked into a small restaurant the other day and was waiting for an order, a man sitting at an adjacent table remarked that "It looks like you have the same thing I have." I did not realize what he was talking about until he said, "CMT," and tapped on the plastic brace on his calf. He is the first person, outside of a doctor, to identify my CMT. I was amazed!

As previously stated, my life has been great and I don't know if it would have been any different if I had not had the CMT. Even though I'm retired, I represent a company in the state of Texas and I travel to meetings in the state, reporting to the President of the company in Boulder, CO. My wife and I have an RV and travel to many states.

I didn't mention it, but to the best of my knowledge, I have never had another pain as a result of my CMT.

—S.C. Mesquite, TX

Dear CMTA,

I would like to make a donation in honor of Stephanie DiCara and her family. Her father is my attorney and has helped me in so many ways that it is only fitting that I repay, in some way, his kindness.

Stephanie is a brave young girl, and her spirit and attitude are wonderful. She is lucky to have a supportive mother, father, and sister. They are a wonderful family. I only hope you find a cure for this disease soon.

I have given to many organizations, but never to one where I actually know someone afflicted with the disease. This will be a regular donation for me!

—A.D. Algonquin, IL

Dear CMTA,

The Philadelphia conference was very informative and all the speakers were just great in trying to answer our questions in “lay-person” language.

I don’t know if anyone has spoken to you since the conference, but we had an interesting experience when we tried to get back to our hotel. We had left our car there and took a cab to the BRB Auditorium, expecting to take a cab back to retrieve our car.

We used a pay phone to call and there was no answer, so we called our hotel and asked them to send a cab to the corner of Curie and Osler. After waiting for over half an hour, my daughter again called the hotel, who told her that cab companies will not pick anyone up from a street corner.

In desperation, she asked a man who was just leaving the building if he would give us a lift. He not only did, but with a smile, and couldn’t have been nicer. When we got to the main street, Civic Center Boulevard, he recognized some people walking who had been at the conference and offered them a lift, too. We wound up with 7 or 8 women in the van. I tell you all this because I only know the man’s name, Steve O’Donnell, and I want to make sure he is thanked.

By the way, I have hearing problems. At the conference, I heard most of what was said and my daughter filled me in on what I missed. I was particularly interested in hearing Dr. Oatis and I could hear every word she said. It must have been the pitch of her voice, plus she spoke slowly and distinctly.

Please thank everyone who got this conference together. My daughter is currently waiting for DNA results on her 6-year-old daughter.

—T.E. PA

Dear CMTA,

Physical exercise is good for you. I know that I should do it daily, but my body doesn’t want me to do too much, so I have worked out this program of strenuous activities that do not require physical effort. You are invited to use my program without charge.

1. Beating around the bush
2. Jumping to conclusions
3. Climbing the walls
4. Swallowing my pride
5. Passing the buck
6. Throwing my weight around
7. Dragging my heels
8. Pushing my luck
9. Making mountains out of molehills
10. Hitting the nail on the head
11. Wading through paperwork
12. Bending over backwards
13. Jumping on the bandwagon
14. Balancing the books
15. Running around in circles
16. Eating crow
17. Tooting my own horn
18. Climbing the ladder of success
19. Pulling out all the stops
20. Adding fuel to the fire
21. Opening a can of worms
22. Putting my foot in my mouth
23. Starting the ball rolling
24. Going over the edge
25. Picking up the pieces

Happy exercising!

—C.G. Delray Beach, FL

IN MEMORIUM

The CMTA mourns the loss of one of its former board members, Steven B. Scofield, following a long illness. Our sympathies are extended to his family and friends.

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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.

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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