JULY/AUGUST
2004

ISBN #1067-0181 ✲ Vol. 19, No. 4

THE CMTA Report

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

Charcot-Marie-Tooth Conference in Johnstown, PA

On May 26, 2004, over 100 people attended the CMT conference held at the Holiday Inn in Johnstown, PA. This event was planned and carried out by the Cambria-Somerset Council for the Education of Health Professionals in conjunction with the Conemaugh Health Systems.

After welcoming and introductions by Dr. William DeMayo from Conemaugh and CMTA Executive Director Charles F. Hagins, the conference began with an overview of CMT by Dr. Michael Shy, Wayne State University, Detroit, MI. Dr. Shy began by explaining that CMT bothers patients in a length-dependent manner—feet first, then legs, fingers, and then arms. The longest nerves are the first to be affected. Currently, he said there are 26 distinct forms of CMT found on 44 loci. Within a week of the conference, mitofusin, a new gene that affects the mitocondria and causes CMT 2A had been found.

Many patients question how they can have CMT if neither of their parents had it. Dr. Shy explained that over half of all patients with Type 1B have new mutations that are not inherited from their parents.

The second speaker, Dr. Neil Busis from University of Pittsburgh—Shadyside, discussed, among other things, the two kinds of nerves that relate to people with CMT. Efferent nerves control movement, secretion, and regulation of body temperature. Afferent nerves affect feeling—both conscious and unconscious.

Michael Shy, MD, Patrick Torchia, Barb Duryea, Charles F. Hagins, and Jan Goodard gathered at the dinner Tuesday night before the Johnstown conference to recognize the important contributions to CMT awareness.

Ironically, CMT patients complain most about negative motor symptoms and positive sensory symptoms. Negative motor symptoms include leg weakness and muscle wasting, distal toe and ankle extensor weakness (causing tripping on rugs and uneven ground), and finger weakness (difficulty in opening jars or turning a key in locks). Positive sensory symptoms include prickling, searing, burning, and tight band-like sensations. Neuropathic pain is an extreme example of a positive sensory symptom.

The final presentation before lunch was by Dr. Stephen Conti, Director, Division of Foot and Ankle Surgery, Allegheny General Hospital, Pittsburgh. He

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INSIDE:

CMTA
Charcot-Marie-Tooth Association

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

OUR VISION:
A world without CMT.

EMAIL CMTA AT:
CMTAssoc@aol.com

JOHNSTOWN CONFERENCE
(Continued from page 1)

discussed the major orthopaedic problems of CMT, including scoliosis (curvature of the spine), hip dysplasia (a special form of arthritis in the hips), and foot, ankle, and lower leg problems such as foot drop, high arches, and a loss of feeling.

In the embryo, the foot begins as a single block of tissue. While in the womb, the foot develops all the necessary structures, (bones, muscle, tendons, and nerves) that will exist in the adult foot. In CMT, the child is born with a normal-appearing foot. As the child grows, the foot becomes unbalanced due to abnormal muscle forces. Once the child has finished growing and the foot has assumed a high-arched shape, it will remain in that position forever. With time, the high-arch can cause other problems, including clawtoes, metatarsalgia (pain in the bones of the arch, usually worsening with weight bearing), and ankle instability.

Orthopaedic treatment of foot problems in CMT usually includes orthotics (arch supports), bracing to prevent ankle instability, and surgery. The goals of surgery are to balance the muscles, lower the arch, straighten the toes, and tighten the ligament on the outside of the ankle. Initial surgery is best if done early. Then there is a high success rate. Long-term results deteriorate with time from worsening of the neuropa-

News from the Executive Director

On July 8th, over 100 of the world's leading CMT scientists gathered in Antwerp, Belgium, to present their recent research.

Excitement was in the air as Michele Fontes, PhD, Marseille, France, completed his presentation on research using ascorbic acid to stop the progression of demyelination in the nerves of CMT rat models.

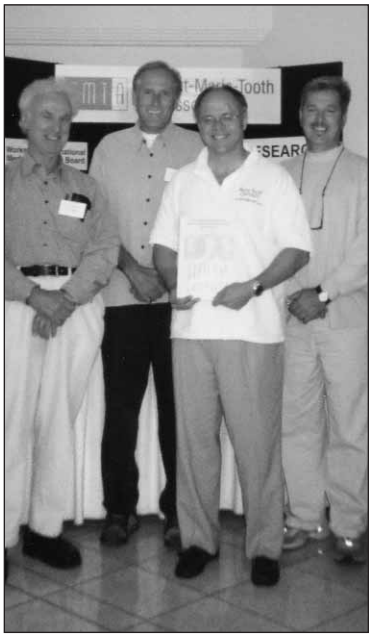
A special session was immediately created to discuss an international collaboration between European and North American scientists, the CMTA, and CMT France and Belgium. It was decided that a special workshop would be convened to develop a protocol for conducting clinical trials on ascorbic acid in Europe and the United States.

Funding for this research will be sought on both continents in a collaborative effort.

There are three benefits to be realized from this effort. First, the potential therapeutic outcome of the research and clinical trials; second, the creation of an organized protocol for future clinical trials; and third, the international collaboration between Europe and the U.S., at an intense level, for CMT research.

I am also very pleased to announce that the House Appropriations Subcommittee on Labor, Health and Human Services, which appropriates money for the National Institutes of Health (NIH) has inserted in the key committee report the following language:

"Charcot-Marie-Tooth disorder (CMT) is one of the most common inherited neurological disorders, affecting approximately one in 3,500 [sic] people in the United States. The Committee is concerned about the prevalence of this disease and its effect on people across the age spectrum and recognizes the value of CMT research for advancing understanding into other neuromuscular disorders. The Committee encourages NIH to identify new research opportunities on CMT that could lead to a relevant program announcement or request for applications.



MAB Chairman Emeritus, Robert E. Lovelace, MD, Co-President/Chairman Patrick Torchia, Current MAB Chairman, Michael Shy, MD, and CMTA Executive Director, Charles F. Hagins represented the CMTA at the consortium in Belgium.

thy and progressive arthritis.

After lunch, the meeting concluded with presentations by Dr. David Riley on alternative medicines in the treatment of CMT and Dr. Robert Chetlin from University of West Virginia on the benefits of an exercise program. The final presenter was Susan Wheeler, BSW, founder of *CMT Today*, who spoke on "Living with CMT" and presented an emotional and dramatic slide presentation on the faces of CMT and hope. ✲

The Committee requests a report on NIH efforts on CMT and CMT-related disorders by March, 2005."

Here's what happens next. The full House Appropriations Committee approves the Labor H Appropriations Bill and associated Committee report (containing our language). That's as far as the language goes in the House. Since it is not part of the bill but only part of the Committee report, we have essentially reached our goal in the House.

We are also concurrently working to place the same language in the Committee report that will be associated with the Senate Labor H Appropriations Bill.

I would like to recognize the work of Jason Steinbaum and Gary Gasper, who played key roles in the insertion of our language in the House report.

This is the first step in increasing NIH funding for CMT research going forward.

—Charles F. Hagins

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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.

Epidemiology of CMT

MICHAEL SHY, MD

(Editor's Note: Several phone calls to the office have centered on the topic of how many people have CMT and what percentage of each type seems to prevail. This excerpt from a presentation by Dr. Michael Shy helps to answer those questions.)

Many of the epidemiologic data on CMT were obtained prior to the era of molecular diagnosis. For example, the prevalence of CMT (all types) was estimated to be 36 per 100,000 in western Norway and 23 per 100,000 in Scandinavia, but only 4.7 per 100,000 in Newcastle-Upon-Tyne. In Olmsted and Wabasha counties in southeastern Minnesota, with a population of approximately 150,000 persons, Dr. Peter Dyck and his colleagues have identified approximately 45 affected persons with CMT1.

In earlier studies, Dyck and Lambert identified 72 of 104 patients with hypertrophic forms and 30 patients with neuronal varieties of inherited neuropathies. In the hypertrophic neuropathy group were 67 patients with a typical CMT peroneal muscular atrophy syndrome and 5 with early-onset Dejerine-Sottas Neuropathy (DSN). In the hypertrophic CMT syndrome group, the disorder was usually inherited as an autosomal dominant trait, but in approximately one fourth of kindreds, the disorder was sporadic or in siblings (the parents having been examined and found to have normal nerve conductions). Syndromes with neuronal varieties included 8 patients with neuronal CMT, 4 patients with distal progressive muscular atrophy, 12 patients with Friedreich's ataxia, and 8 patients with spastic paraplegia.

In their classic review of 227 patients with CMT, Harding and Thomas found that 173 had type I and 54 had type II disorder. Autosomal dominant inheritance could be shown in the majority. In some, autosomal recessive inheritance seemed probable because the disorder occurred in siblings and was not found in patients who were cousins.

With the advent of molecular diagnosis, increasing data are becoming available on the frequency of different genetic types of CMT. The 17p11.2 duplication was identified in 60% to 68% of CMT1 patients in North America and 70.7% of 819 unrelated CMT1 patients in a European collaborative study. CMT X accounts for approximately 10% to 20% of all cases. *MPZ* mutations account for about 5% of patients with type 1 phenotypes, although slightly lower percentages have also been reported. Point mutations in *PMP22* or *EGR2* are less frequent. Remarkably, point mutations have often been identified as de novo or sporadic events without clear family histories. The overall prevalence of HNPP is not known, but 84% of 115 unrelated patients with clinical evidence of HNPP were found to have the chromosome 17p11.2 deletion. Fourteen of 63 families with dominantly inherited neuropathies were found to have features consistent with CMT2 in the studies of Ionasescu, et al. ✲

PFIZER SETTLES NEURONTIN SUIT

(FROM NORD)

Pfizer has agreed to pay a $430 million fine and plead guilty to charges that it illegally marketed the epilepsy drug, Neurontin, for off-label uses that were unproven. Since 2002, the federal government has collected over $2 billion in similar fines from AstraZeneca, TAP Pharmaceuticals, Bayer, and Abbott Laboratories. For the Pfizer case, the illegal marketing occurred at Warner Lambert's Parke-Davis Division. Pfizer bought Warner Lambert in 2000.

Doctors are legally permitted to write prescriptions for off-label uses of drugs, but pharmaceutical companies are not allowed to purposely market drugs for uses that have not been proven. According to the settlement, Parke-Davis "aggressively marketed" Neurontin to treat bipolar disorder, attention-deficit disorder, pain, migraine headaches, ALS (Lou Gehrig's disease), drug and alcohol withdrawal, and restless legs syndrome without proof of effectiveness. A study of bipolar disorder, for example, has shown that Neurontin is as effective as a placebo. An ex-employee in a whistle-blower lawsuit revealed the illegal marketing and provided marketing materials as proof that salespeople were told to sell Neurontin for unproven uses. ✲

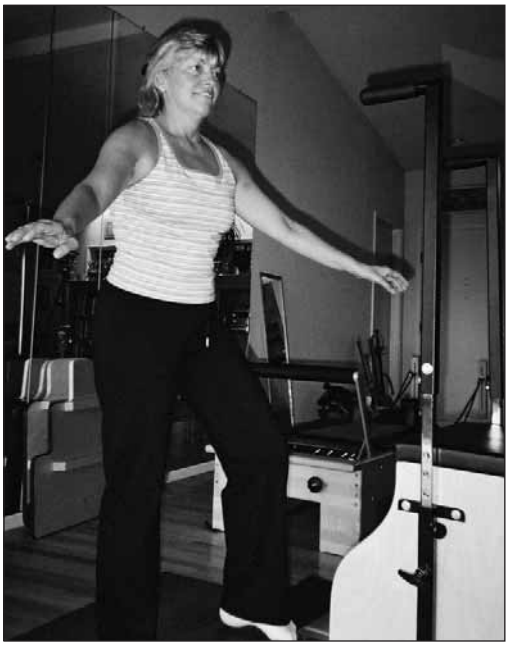
Pilates and Charcot-Marie-Tooth Disease

LAVINIA MAGLIOCOO, PILATES INSTUCTOR WITH
RICHARD GELLMAN, MD, ORTHOPAEDIC SPECIALIST

I had never heard of Charcot-Marie-Tooth (CMT) disease until Nadine, age 51, was referred to me by her physical therapist. Nadine became quite symptomatic at age 42. She suffered from pain in her knees, diagnosed as chondromalacia, a softening of the cartilage on the patella. She also felt weak in her trunk, arms, and hips, in addition to the leg weakness commonly associated with CMT disease. As an adolescent, she described a history of repetitive ankle sprains. Before starting Pilates, Nadine maintained a rigorous exercise schedule that included walking on a treadmill with special shoes to maintain ankle dorsiflexion, and various physical therapy modalities several times per week.

As a Pilates teacher, I am trained to observe how people stand and walk as clues to their muscle imbalance. Because I knew little about CMT, I approached our first session with curiosity and no assumptions. Nadine's stance (standing stability) was poor from her inability to balance and muscle weakness. This caused her to instinctively adopt a "braced" stance in her upper body. It is the protective posture humans adopt when they feel off-balance. Shoulders are raised and drawn into the body and elbows are semi-flexed, ready to break a fall.

Her feet have high, rigid arches and she walked with a footdrop. In order to walk, she must lift her leg higher off the ground with her hip flexors and place the foot on the floor. In testing her muscle strength, her hip flexors and hamstrings were shortened and weak, and she had significant weakness in her gluteal muscles. Her quadriceps were weak and visibly atrophied. Her lower legs had significant atrophy, with no functional strength to lift the foot upward, while her gastrocsoleus muscles were shortened and also weak. The leg muscles that stabilize the side-to-side balance of the foot were also very weak. Her entire abdominal and lower back "core" musculature was weak. I also found her scapular muscles to be weak and unbalanced from the chronic elevation of her scapula, with no scapular rhythm. Scapular rhythm allows smooth motion of the shoulder blade, with no rigidity in how the arms are held.



Nadine works on improving her balance and coordination.

Pilates strengthens the muscles of the core. This includes all the abdominal muscles: rectus abdominis; internal and external obliques; transverse abdominis; iliopsoas, as well as the muscles of the back: multifidi, rotatores, and erector spinae. In Pilates, we emphasize quality and form over quantity. The goal is to develop functional strength, not obvious hyper-musculature. Many exercises recruit subtle muscular responses which develop proprioception (balance) and coordination. This is the prime benefit to people with CMT who have decreased lower extremity proprioception. A Pilates workout can demand awareness, not stamina, and for this reason, it is ideally suited for people with CMT.

PILATES THERAPY

Instruction began on a raised platform called the Cadillac apparatus. Our first sessions focused on retraining her tensed shoulder girdle to allow a more functional scapular rhythm. Her first exercise was an assisted half

(Continued on page 10)

CMT: A Clinical Perspective

ROBERT A. BEDOTTO, PT, CPO, www.ortho-therapy.com

As a physical therapist and an orthotist-prosthetist, I consider myself to be a clinician and rehabilitation specialist. The goal of rehabilitation is to improve function. Treatment must be designed to achieve this goal. Understanding CMT is critical for the clinician and patient alike. Education is the key to understanding. The application of this knowledge to each individual case, along with effort and commitment, will determine the success of the treatment.

Since I have been involved in rehab for 33 years, I treat patients. To a clinician, a CMT patient is also a client. This implies a business relationship with an underlying plan. The client is also a consumer and as such should be aware of the product or service that is being provided. The end result or outcome should be discussed at the beginning, as it is the basis for treatment. Patient care remains critical for success. Individuals must be responsible for their own treatment with the help of a dedicated clinician. Together, we must put the CARE back into health care! Most importantly, my patients are people with a name. In spite of my experience, I remain a perpetual student. I have learned more from patients than I have ever taught them, as the result of good clinical relationships.

I believe in education. Clinicians and patients are blitzed by marketing strategies designed to sell products. We need to separate marketing from science and replace opinions with facts. Patients, physicians, physical therapists, and orthotists must be aware of the need for total treatment. An orthosis (brace) in itself is just a product. It is not a treatment. Treatment begins and ends with physical therapy. Orthotic use and physical therapy cannot be separated. They are both parts of the total treatment.

The fields of genetics and neurology offer the hope of a better future, but what can be done now? Genetic counselors and neurologists do not treat CMT. Technology has offered new hope, but it is not a panacea. We must apply the basic rehab principles of the past to make it work. With this approach, there is something that can be done for CMT today!

> CMT pathology varies significantly among those affected, presenting a unique mechanical profile to the clinician.

DISEASE PROFILE

CMT, like all other pathologies, varies significantly among those affected. While it is extremely important to recognize the presence of CMT early and to understand the pathology, it must be emphasized that each individual presents a unique mechanical profile to the clinician. While there may be similarities, successful rehabilitation lies in recognizing the differences. To treat everyone the same is wrong. Understanding the progressive nature of CMT is paramount to preventing deformity and functional deterioration. While there are clinical needs unique to CMT, treatment in many cases overlaps with other pathologies.

There is no present cure for CMT; therefore, the pathology itself cannot be treated. Deficiencies of the musculo-skeletal system must be addressed. Early detection is critical. Due to the progressive nature of CMT, the causes of potential deformities must be treated early. The complexity of treatment increases as the deficiencies and deformities progress. Preventative treatment is easier than corrective treatment.

The primary causes of deformity in CMT are related to muscle weakness. Left untreated, this weakness will lead to skeletal malalignment. The classic changes in the structure of the feet along with foot drop are symptoms. The primary problem is muscle weakness. Sensory loss, including proprioception (position sense), further complicates the treatment. Secondary problems such as balance, securi-

ty issues, and compensations develop. Ambulation becomes difficult, inefficient, and energy consuming. Gait deviations result.

Many individuals with CMT are treated for "foot-related problems." A foot orthosis or "orthotic" will not address the primary problem of muscle weakness. Too often, this type of treatment continues as the deformities progress. Physical therapists often look at orthotic intervention as a failure of treatment and avoid their use until ambulation becomes difficult or impossible. Orthoses are often used as an afterthought and they are often inappropriate! The result is the same. Deformities progress to the point of "fixation" or contracture. A cycle of regression begins that has nothing to do with the progression of CMT! If the primary problems were treated initially, many of the secondary problems would not develop!

The biomechanics of human gait must be understood to treat the primary problem of muscle weakness. The skeletal system is comprised of levers, with muscle power controlling movement. Weight bearing and gravity affect weak musculature in the stance phase. Orthoses used to prevent "drop foot" solve the symptom in the swing phase only. During stance, these types of orthoses may make walking more difficult! Traditional orthoses incorporate designs and materials that do not address the primary problem. These braces are often discarded since they do not improve efficiency or functional ambulation. It is support in stance that is required. Weak or missing musculature also must be replaced orthotically. The ultimate goal of treatment is to improve the efficiency of ambulation, not just to prevent drop foot.

Another example of treating a symptom is hyperextension of the knee. In the presence of weak knee extensors, the knee hyperextends as a compensatory mechanism. The routine approach is to provide a brace that picks up the foot and forces the knee forward at the same time. In treating the secondary problem, the primary problem has been ignored. This will force the knee to flex in stance, causing more insecurity and increased effort.

TOTAL TREATMENT

An orthosis is not therapeutic in itself. It should be part of an overall treatment plan involving the physician, physical therapist, and orthotist. PTs have a strong biological background, whereas Orthotists have a strong mechanical background. In order to meet the biomechanical needs of CMT patients, total treatment should include the integration of the most appropriate orthosis from the beginning. The physician needs to oversee the total medical treatment for each patient. If proper treatment has been delayed, secondary problems can become primary problems and must be treated separately.

Total treatment begins with a thorough physical therapy assessment including:

- Joint range of motion, including skeletal and muscular factors
- Muscle strength
- Sensory and position sense
- Balance
- Postural evaluation
- Limitations in movement patterns affecting balance and gait
- Full body gait analysis

A complete medical exam should be conducted prior to treatment. Treatment then begins with a discussion of the assessment and the planning of long-term and short-term goals. Depending on the complexity of primary, secondary, and compensatory deficiencies, a specific program can be established. Treatment may consist only of physical therapy at first, followed by the fabrication of an orthosis.

Treatment with the orthosis should include balancing activities and therapeutic exercise in non–weight-bearing postures to emphasize and strengthen appropriate movement patterns necessary for ambulation. Training is necessary to overcome compensations or old habits that may have been acquired over the years. New movement patterns that are ultimately more efficient may feel awkward or more difficult at first. The exercises are designed to enhance appropriate orthotic treatment.

Commitment and effort on the part of the clinician and the patient will lead to improved outcomes if everyone is in agreement with the treatment plan and protocols are followed. Total treatment is not the quick and easy approach, but it is the only way to provide meaningful outcomes. The rehabilitation goal of improved efficiency and function in ambulation ultimately affects one's quality of life. ✻

Remembering Peggy Jones

Peggy Jones of Tennessee was loving and caring in both life and death. Her husband told the CMTA that she always thought of others and wanted her memorial gifts to go to the advancement of CMT awareness.

GIFTS IN MEMORY OF PEGGY JONES WERE MADE BY:

Mr. & Mrs. Dwayne Alley
Ken Bartlett & Family
Mr. & Mrs. John Beddingfield
Ronnie & Ann Butler
James Caplinger
Jimmy Chitwood
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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.

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In honor of (person you wish to honor)

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Send acknowledgment to:

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Occasion (if desired):

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Name:_____

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Amount Enclosed: _____

☐ Check Enclosed ☐ VISA ☐ MasterCard

Card #_____

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Gift Given By:

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William Chase
Louise Potocny

Jasper Kelly
Teena Williams

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Stacey & Jo Bernson

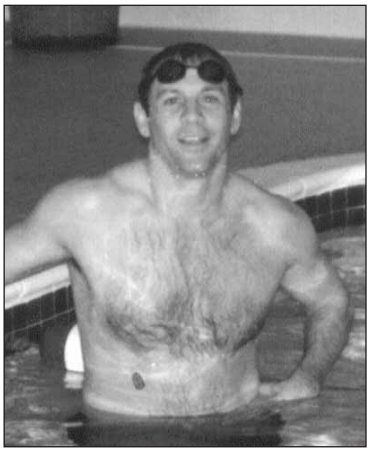
Connie Seagraves
Dorothy Miller

Fundraising News

Congratulations again to Board Co-Chairman and Co-President Steve O'Donnell who completed the swim of the Chesapeake Bay on June 13, 2004, for the third year in a row. Steve's swims have resulted in over a hundred thousand dollars being donated to CMT research.

Betty Chow has organized the second annual Bowl Over CMT to raise money for CMT research. She ran her fundraiser on July 24, 2004, at the Bahama Lanes in Pasadena, CA. She and her husband have spearheaded this campaign of awareness and fundraising.

On July 19, 2004, Board member Robert Kleinman hosted his annual AFA Golf tournament at The Creek, Locust Valley, NY. This is his fifth year of sponsoring the tournament, which netted $36,000 last year and is expected to result in even more research money in 2004.



Steve O'Donnell and other CMTA leaders and members work to raise money for CMT research through swims, golf, and bowling tournaments.

Board Co-Chairman and Co-President, Patrick Torchia, hosted his fourth annual golf tournament at the Sunnehanna Country Club on July 23, 2004, to raise money to create awareness of CMT disorders.

On September 13, 2004, Chris Scarduzzio will host his fourth annual golf tournament in memory of his father John J. Scarduzzio at the Rolling Green Golf Club, Springfield, PA. Chris is the executive chef at Brasserie Perrier in Philadelphia. ✲

PILATES AND CMT

(Continued from page 5)

roll-up. This made her aware of her scapula motion and assisted her in initiating a roll-up. Next, we did a full roll-down and roll-up. The goal was to increase the use of her deeper abdominal muscles. This also helped her gain more mobility in the spine. She had been flexing with all motion coming from her hip joints.

LEG WORK

I began leg work with gluteal and adductor retraining, performing leg lifts while lying on her side. Later, she performed pelvic curls to increase her ability to use her gluteals, learn the concept of pelvic stability, and further strengthen her core.

Nadine, at first, was unable to perform a knee bend on the Reformer, a horizontal sleigh developed by Pilates. As she initiated knee flexion, there was a pattern for her knee to strongly deviate outward. With a ball between her thighs or ankles, we worked on simultaneously recruiting the hip stabilizer muscles. She then began strengthening her quadriceps muscles without knee pain.

Her hamstrings tended to cramp in response to exercise, often requiring a stretch prior to Pilates work. I also stretched her quadriceps and hip flexors, utilizing the Ladder-barrel.

Although previous therapists had worked on gastrocsoleus and toe stretching, I found it extremely helpful during leg-work to massage her legs and feet and then stretch her achilles. Nadine has gained ankle dorsiflexion by stretching out her long-standing achilles contracture from this technique. When we first began, she could not get her heels down. Now, she can do leg work with her toes on the bar and is able to keep her heels down.

We have used the Wunda-chair for many exercises to help strengthen the deep abdominal muscles and the quadriceps. Another exercise focuses on challenging her balance. She faces the chair in a standing position with her foot on the pedal and pushes down.

Nadine performs a knee bend and works to strengthen her quadriceps.

ABS AND UPPER BODY

We continue to focus on her core muscles during all of her Pilates sessions. I found it helpful to have Nadine stand and face a mirror while strengthening her arms and shoulders with a flexible rubber sheet called a TheraBand. The mirror helps Nadine self-correct as she focuses on stabilizing her shoulders. We have progressed to kneeling side arms on the Reformer. This series is especially useful because, as well as working the arms, it offers a greater challenge to balance and stability.

Nadine's fine motor control and strength in her hands have been weakened by CMT. She has difficulty opening jar lids or pulling weeds. Many of these actions can be improved by strength, stability, and control of the entire upper extremity and torso during rotational movements. She has strengthened these muscles with Pilates' exercises as well.

Stretching is extremely important for clients with severe muscle imbalance. Nadine uses a TheraBand to stretch hamstrings. For quadriceps, standing within the frame of the Ladder barrel works very well. We incorporate a pectoral stretch with her standing TheraBand work. Lying supine over the barrel adds an important stretch for her spine and hip flexors.

CONCLUSION

Nadine's balance and core stability are much improved by her Pilates work. She walks at a greater speed and with more ease. These are both quantitative and qualitative improvements whose physical and psychological benefits cannot be overemphasized. Nadine feels more confident. She began Pilates work 6 months ago, coming twice per week. She can now perform an extensive regimen of advanced exercises. A crucial element to our success is Nadine's own spirit. Her courage, sense of adventure, and willingness to experiment has made our work together inspiring, fun, and satisfying. ✲

CMT Support Groups

Bob Budde, Support Group Liaison, 859-255-7471

Arkansas—Northwest Area
Place: Varies, Call for locations
Meeting: Quarterly. Meetings are not regularly scheduled so call ahead.
Contact: Libby Bond, 479-787-6115
Email: charnicoma57@yahoo.com

California—Berkeley Area
Place: Albany Library, Albany, CA
Meeting: Quarterly
Contact: Gail Whitehouse
Email: gwhite@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
Email: pcmobley@mac.com

Colorado—Denver Area
Place: Glory of God Lutheran Church Wheat Ridge
Meeting: Quarterly
Contact: Marilyn Munn Strand, 303-403-8318
Email: mmstrand@aol.com

Kentucky/Southern Indiana/ Southern Ohio
Place: Lexington Public Library, Northside Branch
Meeting: Quarterly
Contact: Martha Hall, 502-695-3338
Email: marteye@mis.net

Massachusetts—Boston Area
Place: Lahey-Hitchcock Clinic, Burlington, MA
Meeting: Call for schedule
Contact: David Prince, 978-667-9008
Email: baseball@ma.ultranet.com

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

Minnesota—Twin Cities
Place: Call for location
Meeting: Quarterly
Contact: Maureen Horton, 651-690-2709
Bill Miller, 763-560-6654
Email: mphorton@qwest.net, wmiller7@msn.com

Mississippi/Louisiana
Place: Baptist Healthplex, 102 Clinton Parkway, Clinton, MS
Meeting: Quarterly
Contact: Flora Jones, 601-825-2258
Email: flojo4@aol.com

Missouri—St. Louis Area
Place: Saint Louis University Hospital
Meeting: Quarterly
Contact: Carole Haislip, 314-644-1664
Email: c.haislip@att.net

New York—Greater New York
Place: NYU Medical Center/ Rusk Institute, 400 E. 34th St.
Meeting: Third Saturday of each month from 1-3 PM of each month
Contact: Dr. David Younger, 212-535-4314, Fax 212-535-6392
Website: www.cmtnyc.org

New York—Horseheads
Place: Horseheads Free Library on Main Street, Horseheads, NY
Meeting: Quarterly
Contact: Angela Piersimoni, 607-562-8823

New York (Westchester County)/ Connecticut (Fairfield)
Place: Blythedale Hospital
Meeting: 3rd Saturday of each month, excluding July & August
Contacts: Beverly Wurzel, 845-783-2815
Eileen Spell, 201-447-2183
Email: cranomat@frontiernet.net espell@optonline.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: Wills Restaurant 405 Wagner Ave, Greenville
Meeting: Fourth Thursday, April–October
Contact: Dot Cain, 937-548-3963
Email: Greenville-Ohio-CMT@woh.rr.com

Ohio—NW Ohio
Place: Medical College of Ohio
Meeting: Quarterly
Contact: Jay Budde, 419-445-2123 (evenings)
Email: jbudde@fm-bank.com

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
Email: jeanie4211@hotmail.com or blzerbabe@aol.com

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

Pennsylvania—Northwestern Area
Place: Blasco Memorial Library
Meeting: Call for information
Contact: Joyce Steinkamp, 814-833-8495
Email: joyceanns@adelphia.net

Pennsylvania—Philadelphia Area
Place: Penn Towers Hotel Conference Room
Meeting: Bimonthly
Contact: Amanda Young, 215-222-6513
Email: stary1@bellatlantic.net

Pennsylvania—State College
Place: Centre County Senior Center
Meeting: Monthly
Contact: Rosalie Bryant
Email: rab296@psu.edu

ASK THE DOCTOR

Dear Doctor,
I have CMT in addition to Crohn's disease. My doctors want to use Remicade intravenously and I am worried that the medication might adversely affect my CMT. Is there a problem?

An MAB member replies:
I am not aware of any toxic neuropathy or worsening of neuropathy following use of infliximab (Remicade). There is one report of visual changes thought to be from optic nerve toxicity, but there is no reason to believe this finding is common or that CMT patients have any increased risk. Other Crohn's disease drugs can worsen neuropathy, including thalidomide and metronidazole and, sometimes, sulfasalazine.

Dear Doctor,
I have been diagnosed with CMT. My condition is deteriorating and I'm having a lot of pain and muscle hardness or severe contractures in my hands and arms. I've tried most of the general medications, such as Neurotin or Elavil, without much relief. Are there any new medicines that are safe to take that might help me?

The Doctor replies:
That is a difficult question to answer. A lot depends on the type of pain you are having; is it crampy, muscle-type pain; stiff, achy, joint-like bony pain; or more severe nerve-related pain? Different medicines tend to work better for certain types of pain than others. For the nerve pain, I have tried Topamax, Lamictal, Mexitil, Zonegran, Trileptal, and even regular pain medicines like Ultram. For the muscle pain, medicines like baclofen, Zanaflex, and Flexeril are sometimes more effective, and the arthritic pain tends to do best with anti-inflammatory medicines like Naprosyn.

Although I suspect you are frustrated, I encourage you not to give up. Usually, some combination of medicines and physical therapy can make things more tolerable.

Dear Doctor,
I was diagnosed about 14 years ago with CMT. I have had numerous nerve conduction studies on my feet, legs, and arms. I had scoliosis and wore a back brace during my teens. I am hearing impaired and have high arches on both feet. Recently, I had the genetic testing for CMT and the results came back "indeterminate." If my genes are normal, is it still possible for me to have CMT?

The Doctor replies:
Commercial testing is still available for only a limited number of the 24 known mutated genes that cause CMT. Therefore, it does not mean that you don't have CMT if the tests come back negative. However, *indeterminate* is not the same as *negative*; rather, this means that the testing agency is not certain if the results are positive. This can be very confusing for both the patient and the physician.

Dear Doctor,
My wife has CMT and is pregnant. She has an infection called group B streptococcus (GBS) and would normally be treated with intravenous penicillin. We know that this is on the list of drugs to avoid and that in large doses is not recommended. Can other antibiotics be used instead? The infection must be treated because it could be dangerous for the baby.

The Doctor replies:
If GBS needs to be treated, I would suggest either erythromycin or clindamycin as alternatives to penicillin. The danger from penicillin is not great, but there are no citations indicating that either erythromycin or clindamycin ever induce neuropathy.

Dear Doctor,
My 17-year-old son was just diagnosed with CMT. I've read that it does not usually affect the brain. When it does, what are the symptoms? I think my son might be having a problem in that regard.

The Doctor replies:
In rare, usually sporadic cases with no family history, an MRI of the brain will show abnormalities in the white matter, or myelin, of the brain. This is not usually associated with any clinical symptoms, but since these cases are rare, we have little information about long-term prognosis. CMT2X is associated with mental retardation. This is an X-linked form of CMT. There are also extremely rare

cases associated with a birth defect that can be associated with developmental delay. You should discuss this with your son's neurologist to see if an MRI is in order.

Dear Doctor,

I'm a 33-year-old female diagnosed with CMT Type 1A after an exacerbation following pregnancy. I seem to be improving, but want to know if I will make a full recovery or have some permanent problem. I have concerns about numbness, proprioception, circulation, and dry skin.

Will additional pregnancies cause further exacerbation? What else causes exacerbation of CMT?

The Doctor replies:

There is not a lot of information about pregnancy and CMT in the medical literature. What is available is biased toward reporting instances in which a woman with CMT has an exacerbation during pregnancy. Given the frequency of CMT and the rarity of exacerbations, one could come to the conclusion that exacerbations during pregnancy are uncommon. However, this might be incorrect. The best study is from 1993 in which 21 women with CMT were retrospectively asked about their pregnancies. There were numerous problems with the study, including the fact that it predated our ability to genetically test for CMT. Some of the women were undiagnosed before pregnancy and thus we do not know for sure if they really had CMT. Regardless, the study found that 50% of women with CMT had an exacerbation during pregnancy, two thirds had permanent worsening of function even after delivery, and 80% of women with exacerbation during one pregnancy had exacerbation in other pregnancies as well.

The exacerbations were increased weakness, increased problems with balance, and decreased ability to walk. The problems you have with circulation and dry skin are more commonly symptoms of the autonomic nervous system, which can occur with CMT1A. Other things that can exacerbate CMT are drugs, diabetes, vitamin and nutritional deficiencies, alcoholism, and systemic disease, which make for a more severe neuropathy than would be expected if CMT were not present. ✲

1500-Mile Bike Ride Follow-Up

Tony Palumbo, 80-year-old New York man, completed his 1500-mile bicycle ride from Jacksonville, FL, to upstate NY to help raise money for a home for troubled children. Two children carried his 21-speed bike on the final leg of the journey as Tony walked to thunderous applause from his supporters waiting behind the Children's Home in Schenectady.

Despite his CMT, Tony said he had no problems at all completing the journey, even though there were days of rain and a few days with temperatures in the 90s. Tony has lived with CMT for years and cannot walk without leg braces. He has, however, made several trips across North America, often as fundraising efforts.

This trip helped to raise money to build a dormitory for some of the 60 children who live at the Children's Home. The non-profit agency helps children who have been abused or neglected.

Tony told the children about the trip and his love of bicycle riding and summed it all up by saying, "If you want to do something, accomplish something, you CAN do it." ✲

Despite his leg braces and CMT, Tony finished his 1500-mile race to raise money for a children's home.

LETTERS

WRITE TO US!

Pat Dreibelbis, Editor
The CMTA Report
CMTA
2700 Chestnut Pkwy.
Chester, PA 19013
or
CMTAssoc@aol.com

The CMTA reserves the right to edit letters for space.

Dear CMTA,

My name is Archit Khanjua and I am a member of the CMT Association. Currently, I am a sophomore in high school. I was diagnosed with CMT type 1A when I was about two years old. With the exception of some limitations, I have managed to live a normal and healthy life. I have never let my limitations block the way of my successes, achievements, and development into adulthood. This was possible because of an association that cares (that's you, guys), parents that always encourage me, and God, who is always there blessing me with strength and happiness.

One recent achievement that I have made, one of my greatest achievements so far, has made my parents, my community, and myself very proud. One of my deepest desires is to direct and produce movies. For high school, we need to accomplish a certain number of community service hours through projects that benefit our community and our world.

I decided to make a small documentary of the community I currently live in and take much pride in, the Hammocks Community in Miami, Florida. After many hours of hard work and fun, I completed my final project. I presented the documentary video to the Board members of the Hammocks Association. To my surprise, they enjoyed the video and offered to buy it for $500 to promote the community to its residents. Since my project was for the community and a service of charity, I felt it was best to give the money I received where it is needed and will be most appreciated. So, as a "thanks" and as an encouragement to finding a cure soon, I am honored to present the CMTA a donation of $500.

—Archit Khanjua

Dear CMTA,

I have CMT and use AFOs. I have lost my grip and solve most of my "no grip" problems with the adaptation of loops. Using waterproof glue (they could be sewn, also), I've glued loops of shoe lace material to the tops of my socks. The loops should be approximately 2 inches in diameter so that two fingers

Good Grips

Utensils designed with comfort and function in mind

(Editor's Note: We featured tools by this company several years ago when the engineer who developed them for his wife first started marketing them. They remain some of the best and most comfortable kitchen and gardening tools on the market because of their cushioned handles and clever design. They are available at most better kitchen stores and gardening centers or can be bought on-line at Oxo's home page.)

SWIVEL PEELER

The OXO GOOD GRIPS Swivel Peeler is an excellent peeler. The sharp stainless-steel blade glides through even the toughest fruit and vegetable skins with ease. The soft, comfortable handle cushions your hand as you peel, and is non-slip, even when your hands are wet. *Price: $5.99*

SWIVEL SCISSORS

You'll want a pair of OXO GOOD GRIPS Swivel Scissors for your utility drawer. These Scissors feature a soft handle that swivels as you cut to absorb pressure and prevent hand fatigue. Try cutting through thicker items like cor-

will comfortably fit inside the loops. The loops must be on opposite sides of the sock so that your sock will go on straight. I use knee-high socks. It helps at times to use powder on my foot and lower leg to get the sock to slide more easily. I've also adapted, with loops of one form or another, almost everything that needs pulling. Velcro can also be useful.

—R.M., by e-mail

Dear CMTA,

I was diagnosed with CMT 40 years ago, when I was 27. Fortunately, the progress was slow and I was able to raise my children and help with grandkids before it became more debilitating. I am now wearing an AFO on one leg and using a walker to help prevent falls.

For some years there has been a numbness on the bottom of my feet, and it was difficult to feel anything when the doctor tested them. Last July, I began "pool therapy," which is simply doing therapy exercises in a pool. In April, I went in for my yearly testing, and found that there is quite a bit more feeling on the bottom of my feet. In some way, those exercises have reawakened some of the involved nerves. We were quite excited by the discovery and I thought if I shared it with you, more patients with CMT might want to try it.

—M.A., Colorado

Dear CMTA,

I just want to express my deep sadness and sorrow over the death of Ann Lee Beyer. I only met her once, but was impressed by her dedication to our cause. She seemed, to me, to be a genuinely friendly and outgoing person. I had no idea she had another health problem besides the CMT that many of us share. I'm going to miss her now that she is no longer with us. My condolences go out to her family and our CMT family. She will be missed.

—W.S., Kentucky

Dear CMTA,

Please accept this donation from my 13-year-old daughter, Stephanie, in honor of her bat mitzvah. She would like it to go toward the research into Charcot-Marie-Tooth, which has affected a close friend of mine. We hope this donation will be helpful.

—L.H., Maryland

Editor's note: The donation totaled $218 and, combined with the $500 gift from Archit, will help in funding a research grant. More than that, the two gifts demonstrate the generosity and "right" thinking of our young people.

rugated cardboard or coarse twine; the swiveling handle ensures that the Scissors are comfortable, even during these tougher tasks. Spring-loaded for easy cutting, the Scissors also lock shut for safety. *Price: $19.99*

GEL-E GARDEN ESSENTIALS SET

The OXO GOOD GRIPS Gel-e Garden Essentials Set contains three popular and necessary tools for planting and maintaining a lush garden. The Gel-e Trowel, Gel-e Transplanting Trowel, and Cultivator are all constructed of high-quality stainless steel for strength and durability. The Gel-e Trowel, the most versatile garden hand tool, can be used for digging and shaping holes, scooping or smoothing soil, and even up-close weeding. The Cultivator is ideal for loosening and preparing soil, pulling up young weeds, blending compost or fertilizers into soil, and aerating to make watering more efficient. The Gel-e Transplanting Trowel is perfect for digging deep, narrow holes to plant seedlings and for easily lifting out root balls without damaging the plant or its surroundings. *Price: $24.99*

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

CMT PATIENT MEDICATION ALERT:

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

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

Uncertain or minor risk:
5-Fluouracil
Adriamycin
Almitrine (not in US)
Chloroquine
Cytarabine (high dose)
Ethambutol
Etoposide (VP-16)
Fluoroquinolones
Gemcitabine
Griseofulvin
Hexamethylmelamine
Hydralazine
Ifosfamide
Isoniazid (INH)
Mefloquine
Penicillamine
Phenytoin (Dilantin)
Podophyllin resin
Sertraline (Zoloft)
Statins
Tacrolimus (FK506, Prograf)
Zimeldine (not in U.S.)
a-Interferon

Negligible or doubtful risk
Allopurinol
Amitriptyline
Chloramphenicol
Chlorprothixene
Cimetidine
Clioquinol
Clofibrate
Cyclosporin A
Enalapril
Glutethimide
Lithium
Phenelzine
Propafenone
Sulfonamides
Sulfasalazine

What is CMT?

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

THE CMTA Report

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