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The CMTA Report

APRIL-MAY 1999

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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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The Armington Research Challenge 1999 *Take It Personally—Then Take It Beyond*

Research generates hope; hope that cutting-edge technology may one day lead to treatment for our children and grandchildren. With the help of the Evenor Armington Foundation, the CMTA has greatly increased its commitment to funding CMT research. Begun in 1996 as a \$25,000 one-to-one challenge, the terms were increased to a 3:1 arrangement which requires the CMTA to raise \$150,000 to receive a \$50,000 matching grant from the Armington Foundation. Having successfully achieved last year's goal, more than \$340,000 has been raised for CMT research in three years. In this final year of the Evenor Armington Foundation Research Challenge, we must raise nearly \$100,000 before June 30th to qualify for matching funds.

"Take it personally" has been the rallying cry, but this year we must go beyond an individual focus to engage the support of others, too. The effects of CMT often extend beyond the affected individual, impacting his or her entire circle of acquaintance: friends, family and associates. There is a way to generate support within our own circle to meet the Armington Challenge.

Several families, most notably, the DiCara Family of Illinois, have reached out to their friends, families and associates to support the CMTA through a personal appeal. This is a

Please help us advance research with a gift before June 30.

highly effective way to "take the Armington Challenge personally," take it beyond oneself and take the message of CMT and its research into the broader community. Other families are getting involved.

New CMTA board member Carol Henderson is rallying her family's support for CMT research. "Challenging" someone is merely asking them to support something very important: CMT research. Whether it's one person or ten people, we need to broaden our outreach and multiply the results.

Since this is the final year of the Armington Challenge, it is critical that we expand the base of donors to insure that the research funding momentum continues for years to come. Leadership supporters like the Buuck Family, the Wechslers, Ross and Cody Jones, Richard Brunetti, Rick Alber and others helped insure last year's success. To their leadership, we must add greater participation and increased giving from all members and friends. To be successful, more individuals and families are needed to "take it personally—then take it beyond."



New CMTA board member Carol Henderson (far left) with her family: Mr. and Mrs. Henderson, Bernadette Lewis (center) and Annie Crowwell (far right). Spurred by the Armington Challenge, Carol is rallying her family's support for CMT research.

Begin with Flynn

By Paul R. Flynn, Executive Director

Are you willing to enlist the support of friends and family to benefit the CMTA? Don't know where to start? To learn how, please call the CMTA office at 1-800-606-2682 or check our website (www.charcot-marie-tooth.org). We would love to help you help us!

"Our prayers are answered not when we are given what we ask, but when we are challenged to be what we can be."

—Morris Adler

The Armington Research Challenge began not with a prayer but with an invitation. In 1995, the CMTA Board of Directors received a letter in which the Armington Family presented a wonderful opportunity—a challenge to raise more money for CMT research. At the time, the offer to receive a \$25,000 matching grant in exchange for raising \$25,000 from our members seemed both exciting and "reasonable." Though we had not previously raised that much money, the rationale was, "...surely CMTA members will respond generously to such an appeal..." Did you ever!

In the first two years of the Armington Research Challenge, CMTA members nearly doubled the \$25,000 amount required to receive the matching grant; almost \$50,000 was given to the research fund in 1996 and 1997. That resounding success indicated a capacity

and willingness of CMTA members to do more, prompting the Board of Directors to "renegotiate" the challenge to its present terms. The Evenor Armington Foundation will provide a \$50,000 grant if the CMTA matches it 3:1, raising \$150,000 for research. It was, and remains, an ambitious proposition, a significant commitment to advancing CMT research. Last year's success has set the stage for this final year of the Armington Challenge...

Success is not a foregone conclusion.

Achieving our goal this year will require more people to give for the first time and prior supporters to stretch further, to do more. As is mentioned in the cover story of this issue, we must take the Armington Challenge personally, "then take it beyond oneself and take the message of CMT research into the broader community." From its inception, I have viewed the Armington Challenge as more than an opportunity to increase funding for research, valuable as that is. It has been a catalyst, a context for organizational growth and as Adler's quotation above suggests, "a challenge to be what we can be."

NOTES IN BRIEF...

Westchester Broadway Theater Benefit: For the second year, the Dreamcoat Singers and George Puello of the Westchester Broadway Theater (WBT) welcomed us to their "home" and allowed the CMTA to share the stage for a benefit performance. The show, "Songs of the Century," featured the talents of kids aged 9-16 who danced and sang show tunes from "Miss Saigon," "Grease," "Annie," and others. The partnership with the WBT has been a wonderful way to generate funds and raise awareness about Charcot-Marie-Tooth disorders. Additionally, our friends at WHUD radio in Peekskill, NY helped us again with promotional messages and ticket giveaways. Special credit must be given to event co-chairs Nancy Homyak and Lawrence E. Gomez for their dedication and hard work on another successful event. Many thanks to Nancy and Larry and all who supported the event!

The 2nd World Congress in Neurological Rehabilitation: Chairman of the Board, Ann Lee Beyer and President Jack Walfish participated in The 2nd World Congress in Neurological Rehabilitation held in Toronto, Ontario, Canada from April 14-17th. This was the first time the CMTA has been represented at these meetings which focus on the rehabilitation aspect of neurology. One presentation of particular interest to our members was on "Neuropathies: Promoting Rehabilitation Through Regeneration," presented by a member of our Medical Advisory Board, Steve Scherer, MD, PhD, from the University of Pennsylvania focusing on the molecular biology of myelin.

Perhaps because this was the first time the CMTA had participated in this Congress with an information booth, there were far too many visitors—doctors, physiatrists and therapists—who seemed very surprised to learn that there is an organization concerned only with CMT disorders. They know us now but the experience suggests that we still have a major Public Relations job to do!



SPECIAL OFFER: Get All The Facts...

For the first time, you can get the entire CMT Facts Series (I, II, III, and IV) for the low price of \$16.00, plus \$3.00 for shipping and handling (see the order form at right). **This is a special offer being made to "active" dues-paying members of the CMTA.**

Purchased separately, the CMT Facts Series would cost \$21.00 for active members—that's a \$5.00 savings for the whole set. Please note, only Facts I and II are available in Spanish.

Do you already have one or two issues in the series? Why not buy the full set and share those you already have with family members? You might consider giving a set to your doctors or making the series available to a local clinic or library to help raise awareness about CMT disorders.

The newest to the series, *Facts IV* is 32 pages in length with six different subject areas including: Medical Information and Research, CMT Survey Results, Living with CMT, Children and CMT, Legal Information, and Q&A.



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NEW HOPE FOR GENE THERAPY

From Alliance Alert, a publication of the Alliance of Genetic Support Groups.

Researchers at Duke University, led by geneticist Andrea Amalfitano, reported in *Human Gene Therapy* that they have developed a new viral vector that can evade the immune system. Successful gene therapy depends on replacing a non-working gene with a working gene in the cells in the affected organ or system. To do this, many researchers are using viruses that are modified so that they cannot make people sick. These viral "vectors" carry functioning copies of human genes, which are added to the viral DNA by researchers. Once the virus gets to the human target cells, the virus inserts the working copy of the gene into the target cell's genome, thus allowing the cell to use the working copy of the gene. However, the vectors currently used in gene therapy are recognized and attacked by the human immune system. Once the immune system attacks the virus, both the vectors and the cells into which they have inserted the gene are destroyed. If these new vectors work as planned, a major obstacle to gene therapy could be avoided.



CMTA 1998 RES

Summary reports from three researchers

Summer Grant Review: Efficacy of Conservative Support and/or Realignment-Based Therapies for Managing the Charcot-Marie-Tooth Patient

By **CHANDA J. WAN, BA, HOWARD HILLSTROM, PhD, JAMES MCGUIRE, PT, DPM, KENDRICK WHITNEY, DPM**

[Editor's note: Chanda Wan is a second year student at Temple University School of Podiatric Medicine and the grant recipient of one of the CMTA summer student fellowships.]

Charcot-Marie-Tooth (CMT) affects about 150,000 people in the United States alone, making it one of the most common hereditary disorders. The serious effects associated with this neuropathy include: peripheral muscle weakness, foot and hip deformities, pain and ulcers, gait abnormalities, and scoliosis. CMT patients have traditionally been given supportive type therapies such as molded ankle foot orthoses (MAFO)¹ for their lower extremity muscle weakness or custom molded in-shoe neutral-position foot orthoses (NPFO)² for their foot and ankle malalignments. It is not clear which conservative support and/or realignment therapy yields the best clinical outcome for managing the CMT patient. Therefore, this pilot

study was conducted to evaluate objectively the efficacy of MAFOs with and without integral NPFOs for improving biomechanical function.

A repeated measure design was implemented comprising four CMT patients with no previous history of foot and ankle surgery. Each patient was provided with three treatment approaches: 1) a custom MAFO, 2) a custom molded NPFO, 3) a custom molded NPFO that has been integrated with the MAFO. These treatment groups represented therapies providing: 1) support, 2) realignment, and 3) support and realignment. Clinical and objective gait parameters in addition to patient self-assessments were employed to determine clinical outcome.

The results of the study indicate that self-selected walking speed, the temporal sequence of loading, center of pressure excursion index (CPEI), step length, stride length, and pelvic tilt angle improved with treatment with MAFOs and NPFOs integrated with MAFOs. However, a larger sample size is needed to improve the statistical power for comparisons between the MAFO and the NPFO integrated with MAFO (combination of the two) treatment strategies.

Armington and Buuck Review from Wayne State

By **AGNES JANI, MD**

I am writing to submit a Progress Report to the Charcot-Marie-Tooth Association for the Armington and Buuck fellowships which I have received from the CMTA for the past two years. My work has focused on the development of a gene therapy system utilizing adenoviral vectors to introduce genes into the peripheral nervous system (PNS). Because of the support of the CMTA, I have been able to make substantial progress in these studies.

In the March issue of *Human Gene Therapy* (Vol 10:787-800) we have a manuscript, of which I am the first author, entitled "Modulation of Cell-mediated Immunity Prolongs Adenoviral-mediated Gene Expression in Sciatic Nerve." I am also the first author on a manuscript in press in the *Annals of the New York*

¹ MAFO: A lightweight plastic brace that provides extra support for the posterior calf, ankle and foot structures. It is the most common conservative intervention applied to CMT patients. Typically, a MAFO is flat in the plantar region, and hence does not address the issue of foot and ankle malalignment.

² NPFO: Neutral-position foot orthoses supply medial longitudinal arch support and correct for varus or valgus deformities within the hindfoot and forefoot but do not offer direct support for weakened muscles.

RESEARCH REVIEWS

who received CMTA grants in 1998.

Academy of Sciences, entitled "Overcoming Cellular Immunity to Prolong Adenoviral-mediated Gene Expression in Sciatic Nerve." In both of these manuscripts, we demonstrate our ability to not only introduce genes with adenoviral vectors in Schwann cells *in vivo*, but also, by suppressing the host immune response, we showed that we can continue to express the genes we are interested in for months in the nerve.

As part of my fellowship, I also worked to rescue nerves in P₀ knockout mice with P₀-expressing adenoviruses. P₀ knockout mice are animal models for CMT 1B. In these studies we were able to introduce the P₀ gene into the mice with a P₀-expressing adenoviral vector. However, we were never able to introduce enough P₀ protein to reverse the peripheral neuropathy. These results were presented at the Third International Conference on CMT disorders this past October in Quebec, as well as at the American Academy of Neurology Annual Meeting in Boston, 1997.

Our group, led by Drs. Shy and Kamholz, has become increasingly aware that even in demyelinating forms of CMT, weakness and loss of sensation are caused by secondary damage to the nerve itself, or the axon, rather than by damage to the myelin where the initial damage to the nerve occurs. As a result, we are changing slightly the focus of our gene therapy studies so that we can introduce genes for growth factors into nerves to prevent the degeneration of the axon. I will be presenting our preliminary studies in this area at the upcoming American Academy of Neurology meeting in Toronto, Canada, in April. In all of these manuscripts and presentations, I have gratefully acknowledged the support of the Charcot-Marie-Tooth Association, as well as the support of the Armington and Buuck families. As soon as we have reprints, we will make copies available. Without the support of all of your members, I would not have been able to conduct any of this research over the past two years. It has been an inspiring experience for me. I have now been accepted into the Neurology Residency program at Wayne State University. Thus, I will become an independent neurologist conducting research into gene therapy for Charcot-Marie-Tooth disease in my future professional life, largely because of your support and the support of my mentors.

Thank you again for the opportunity you have given me. I will continue to think of myself as a fellow of the CMTA.

Armington Fellowship Review from Baylor

By LAWRENCE REITER, MD

Charcot-Marie-Tooth disease (CMT) is a heterogeneous group of disorders that affect the peripheral nervous system. Several genes have now been identified that can cause a CMT phenotype when mutated or even duplicated. Dr. Lupski's laboratory, where my fellowship was performed, has worked extensively on determining which mutations in the peripheral myelin protein 22 (PMP22), myelin protein zero (MPZ), early growth response 2 (EGR2), and connexin 32 (Cx32) genes can cause CMT. His lab was also instrumental in demonstrating that three copies of the PMP22 gene acquired through DNA duplication can also result in CMT. Despite these advances, however, genetic linkage analysis of families with CMT indicates that there are still at least seven other yet unknown genes that can also cause CMT. My fellowship project was to attempt to identify some of these genes using a DNA sequencing approach. We know that the genes involved in CMT are expressed in the peripheral nerve myelin and that the majority of those identified thus far that cause CMT are integral membrane proteins, which span the phospholipid bilayer of the myelinating Schwann cell. Therefore, our approach was to construct a peripheral nerve expressed sequence tag (EST) library to identify the additional CMT genes.

After several months, we were able to obtain a fresh sample of human sciatic nerve tissue to prepare messenger RNA for construction of the library. I was able to obtain several micrograms of pure peripheral nerve mRNA from these tissue samples. This mRNA was then used to construct more stable complementary DNA (cDNA) for the library. A library was successfully constructed containing insert sizes ranging from 1

continued on page 6

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continued from page 5

kilobase to 6 kilobases (for reference the cDNA for PMP22 is approximately 2 kilobases long). Our plan was to sequence 600 cDNAs from this library and use several computer programs to identify those cDNAs encoding proteins that contain transmembrane domains like the known CMT genes. Unfortunately, we experienced technical difficulties with the vector and host bacterial strain of the library. We were able to sequence several cDNAs from the library, however, and found some of the genes we expected, like PMP22 and MPZ. Another member of the laboratory is now trying to subclone the cDNA library into a different vector so that our sequencing goal can be accomplished.

During my fellowship period I have also been involved in two other CMT-related projects. One project involved searching the human genome for the locations of traveling bits of DNA called mariner elements. My work in Dr. Lupski's laboratory had previously implicated these elements in the generation of the commonly inherited CMT1A duplication. We were interested in identifying other mariner elements in the human genome to see if they were located at other sites where homologous recombination can cause human genetic disease. We were able to identify 109 of these elements located on all human chromosomes but the Y chromosome.

The locations of these elements actually did coincide with the locations of 12 known genomic disorders, or diseases that result from rearrangements mediated by region-specific low-copy DNA repeats (like CMT1A-REP). This work is currently in review for publication.

The other project I was involved in was the analysis of DNA sequence from within the CMT1A duplication. Our group and others have predicted that as many as 30 genes may lie in the 1.5-megabase region duplicated in patients with CMT1A. We want to know what types of genes are in this region for two reasons: 1) if we can understand what types of genes are not dosage sensitive we may be able to better understand why genes like PMP22 are dosage sensitive; and 2) some patients with the CMT1A duplication have subtle phenotypes in addition to peripheral neuropathy and the genes located within the duplication may be involved in generating these additional phenotypes.

Finally, I would like to thank the CMT Association for allowing me to extend my work on CMT in Dr. Lupski's laboratory. Although I am now moving on to a different genetic system (*Drosophila melanogaster*), I will forever be indebted to the members of the CMT consortium of laboratories. I am especially grateful to my former thesis advisor Dr. James R. Lupski and our collaborators Dr. Christine Van Broekhoven in Belgium and Dr. Bernd Rautenstrauss in Germany.

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, 601 Upland Ave., Upland, PA 19015.

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Health Care Professionals: Who Does What in the Diagnosis and Treatment of CMT

Primary Care Physicians: This doctor was formally called the “family doctor.” Rather than specializing in one aspect of medical care, this doctor specializes in the care of the whole person. In the HMO concept of health care, you must see the primary care physician first to get a referral to a specialist. Primary care physicians can be internists, family practitioners or pediatricians.

Podiatrists: If your problems first manifested as hammer toes or bunions or calluses, it’s possible that a podiatrist first mentioned CMT to you. A podiatrist is a specialist in diagnosing, treating and preventing diseases and malfunctions of the foot. Podiatrists are licensed to prescribe medicine and perform surgery. They are familiar with foot problems, including gait disorders in children, ankle injuries, bunions and hammer toes, and the care of foot ulcers, toenails and infections.

Neurologists: Neurologists specialize in the nervous system and the muscles. They are particularly interested in the diagnosis of neuromuscular diseases and it was probably a neurologist who first definitely diagnosed you with CMT. They may have run nerve conduction velocities (NCV) or electromyograms (EMG) on you in the course of diagnosing your CMT. They can prescribe drugs to treat your symptoms, such as pain medications and muscle relaxants.

Physiatrists: This speciality is newer than the others. A physiatrist is a doctor of physical medicine and rehabilitation. They help people cope with the physical aspects of a disorder such as movement corrections and strength and flexibility issues. They are highly skilled in the areas of exercise and equipment use. They frequently oversee the departments of physical therapy and occupational therapy.

Physical and Occupational Therapists: Physical therapists deal with the large muscles and with physical strength and endurance while occupational therapists deal more with the small muscles and the ability to perform daily tasks. In simple terms, physical therapists often deal with issues of gait abnormalities and walking

problems and occupational therapists may design exercises to keep flexibility and strength in the hands and arms. Both specialists know about aids for daily living and gadgets that can make life easier. Both specialists also can design exercise programs for persons with CMT to follow at home.

Orthopaedists: If you’ve had serious corrective surgery, you have probably met an orthopaedic surgeon. These doctors specialize in bones, joints and their associated structures, such as muscles and tendons. Many young patients with CMT have had tendon transfers, tendon lengthening, and procedures to lower the arch and straighten the toes. Orthopaedic surgeons perform corrective surgery to help patients avoid the complications of foot deformities and gait irregularities.

Genetic Counselors: These specialists combine counseling skills with an understanding of the medical and scientific aspects of genetics. They can coordinate genetic testing and can provide families with an understanding of their inheritance pattern or how the disease has been handed down in their family. Genetic counselors are often contacted when a couple begins to plan a family. They can provide supportive guidance with regard to genetic disorders like CMT and the likelihood of passing the disorder to a new generation.

Social Workers: Medical social workers are found in hospitals and clinics and generally their speciality is coping with illness and disability with practical solutions such as insurance reimbursement, equipment, housing needs and home care. Clinical social workers, on the other hand, function more as counselors dealing with the psychosocial issues of disability.

Psychiatrists: Problems associated with physical disability can cause psychological distress, so you may be referred to a psychiatrist at some time. These specialists are medical doctors and can prescribe medicines such as antidepressants. They will counsel patients and families and help people put problems in a more helpful perspective.

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SURPRISE ATTACK

The Gentle Art of Fighting Back

By **CAROL SOWELL**

Crime and danger are rampant, and everyone's a potential victim. Some predators think people with disabilities are the easiest victims of all. Some predators have a surprise in store.

Self-defense courses teach people with any level of ability to ward off personal attack by knowing how to use everything available as a weapon—from arms and legs to car keys or a wheelchair's wheels. Using such techniques can enable intended victims to:

- avoid suffering serious injury or losing consciousness by deflecting punches or breaking the attacker's balance and thwarting his intention
- surprise an attacker so that he gives up and leaves
- gain enough time for someone else to come to their aid
- immobilize the attacker and end the fight quickly

Self-defense has been learned by people with cognitive, visual and hearing disabilities, as well as by amputees and wheelchair users who have strong arms and those who don't. For the latter, the chin and head are weapons, as are the teeth and the wheelchair itself.

Almost all self-defense courses draw on the Asian martial arts, all of which combine psycho-

logical, spiritual and physical lessons. Along with techniques for disabling an opponent, students learn alertness, breathing techniques, concentration and confidence.

Bill Austin of Perry, Okla., has studied martial arts for 11 years and earned a third-degree black belt in a style called Wu Shu Kung Fu, which combines techniques from several of the arts. It focuses on "fighting inside" or "short techniques." Rather than the showy kicks and leaps of competitive martial arts, masters of this technique are prepared to fight up close.

Austin, who uses a power wheelchair because of spinal muscular atrophy, has defended himself physically. He's prepared to respond to an attacker by "leaning back and then going forward with my hand on my electric knob for my wheelchair. If they get close to me I'd bite them, or hit them with my head, shoulder, whatever to draw them in." A power wheelchair slamming into the shins is a serious deterrent.

"A lot of it is surprise, and a lot of it is you have to put yourself in a position to react normally without any glitches, and that's where concentration and relaxation come in," Austin says.

P.J. Dixon of Tucson, Ariz., studies a technique called Taijutsu, based on an ancient ninja skill of fighting with the hands. Dixon, who has Charcot-Marie-Tooth disease, is helping his

Left to right: P.J. Dixon defends himself against a "front strangler" attack from the much larger, stronger David Fry. By turning his head and shoulders, he escapes Fry's grip, then moves his wheelchair forward and catches the attacker's leg, while immobilizing his arm with his chin movement. Dixon finishes the technique by hitting Fry's kidneys with the hard part of his skull.



teacher, Jeffrey Prather, instruct classes for people with disabilities.

In Taijutsu, Prather says, "We use timing and distance, instead of speed and strength." While one of Prather's wheelchair-user students, LeNae Liebetau, can perform many techniques with her hands and arms, Dixon relies on his head, chin and quick wheelchair maneuvering for self-defense.

Liebetau says, "I feel better knowing there's something I can do, things I can try." Because an attacker is committed to moving in one direction, self-defense instruction focuses on knowing how to throw him off balance at the right time. Liebetau says, "You learn how to work their body to your advantage." Dixon, a third-degree black belt, values the spiritual dimensions of his martial arts training, which has helped him handle many situations in life.

"The objective is to recognize what you're capable of. It changes your perspective on life. It's a way of understanding how things work in the universe, and incorporating wisdom and knowledge in all areas."

Last year, Dixon's wheelchair was hit by a car turning into traffic. "I remained calm and moved with it," he recalls. "I was not afraid. I tucked and rolled to get out of the way of the wheel."

He attributes his calmness to his self-defense training. "Nothing got broken but the wheelchair. I reacted appropriately. But being clear-minded helped." If you'd like to learn self-defense, look for a class that's geared toward people with disabilities. If there isn't one in your community, visit some classes and talk to instructors. You need one who's willing to work with you to modify techniques and adapt to your needs.



Left: Taijutsu instructor Jeffrey Prather simulates an attack with a crowbar. P.J. Dixon traps Prather's arm with his chin and his foot with his wheelchair. The attacker, experiencing pain in three spots, will either loosen his grip and lose his balance or get a broken arm.

Liebetau cautions, "Know your body and your limits, how much activity you can handle. Part of learning self-defense is to advocate for yourself and communicate to others."

This article appeared in Quest, Volume 6, Number 1, 1999, the publication of the Muscular Dystrophy Association. It is reprinted with the permission of Bob Mackle, the editor of Quest.



IDEAS FROM MEMBERS

One member in NY recommends the use of an air mattress for CMT patients with backache problems. He says he has never slept as well or awakened with so little pain since getting the air bed.

Another gentleman reader suggests that men who have problems pulling their pants over their leg braces use plastic bags over their legs to make that task easier. Another man who uses leg braces suggests putting the slacks on first and then putting on the braces to avoid the issue of pants catching on the braces.



Why My Pain Is Not Your Pain

from *The Culture of Pain*

By **DAVID B. MORRIS**
(1991, University of California Press)

[Editor's note: The following excerpts are from an article that David Morris wrote for the Arthritis Today magazine. The concept that he articulates in his book and his article is that the "myth of two pains" (that mental pain and physical pain are separate) is inaccurate.]

Although pain often makes us feel isolated and uncommunicative, we share the underlying biology of pain with other humans. Neurons that specialize in pain are standard equipment in all mammals. Despite our shared biology, one truism seems undeniable. My pain is not your pain, and your pain is not mine. We come to pain, like love, from very different angles. Pain is always personal and always subjective.

Most of us pick up whatever we know about pain from the popular culture, where information is often inaccurate and outdated. People who suffer from chronic pain know pain intimately; still they have reason to increase their knowledge. You might say that it's the doctor's job, not yours, to know about pain. Doctors, however, get far less education in pain than most patients assume. In 1987, John Liebeskind, PhD, stated that the students he taught at the UCLA School of Medicine received just three hours of lectures on pain treatment during their four years of medical study.

Medical education has changed since 1987, but not enough. Medical students, today, are taught about the physiology of the human pain-transmission and pain-modulation systems; however, a full understanding of pain demands that we consider issues that don't easily fit into a science-based, biomedical curriculum—issues that involve the complexities of culture and individual perception.

Pain, it turns out, is not simply a matter of nerves and neurotransmitters. When a pain signal reaches the brain, it is meshed with all experience stored there and it is altered in a way that is unique to that particular person's moment in life. Emotional states also influence whether the pain stimulus is perceived and how. For this reason, specialists now insist that pain is not a sensation, but a perception. It is a

complex experience influenced by many of the forces that shape our lives: social practices, interpersonal relations, psychology, upbringing, heritage, work circumstances, personality, gender and age.

In effect, pain exists only as we perceive it and we perceive it through the filter of human experience. The new thinking about pain sees it neither solely as biological nor solely cultural, but mixed. Pain is not something objective, like a virus, but always subjective and always a psychological state. We know, now, for instance, that in couvade syndrome, a male partner of a pregnant woman can experience many symptoms of pregnancy, including pain. We know that "secondary gain" such as a more attentive spouse or a disability check can prolong pain and complicate treatment. We know that emotions, especially fear and anger, can intensify pain. We know that people close to us can make a difference in the pain we feel. Negative responses from family members and close friends have been shown to increase the severity of pain in patients with chronic pain. We know that a diagnosis or a lack of one can influence pain. We know that personality, as influenced by heritage and upbringing, can make some people wince and gripe while others insist they feel fine. We know that strong social or professional pressures can lead people to suppress or disguise their chronic pain by cheerfulness, denial or tact. We know that for many people, the most effective source of endurance and support is prayer. The simple belief in help from a higher being seems to make their pain more bearable.

The view that pain is a perception ranks among the most important changes in modern thought. Like other big changes, it will take decades to complete and won't come easily. The most destructive myth still in practice is "It's all in your head." There is no such thing as imaginary pain! Pain is always real to the person who experiences it. There is pain without known cause, pain without observable tissue damage, but there is never imaginary pain. Pain is always a joint creation of bodies and minds inextricably embedded in a social and natural world. What matters most is that we recognize that our minds and emotions help to constitute the complex experience we call pain.

Healing Journey Cuts a Path Through Alternative, Traditional Terrain

[This article about newsletter contributor, Cynthia Gracey, appeared in Up Close, a publication of the Upledger Institute. Cynthia has written several articles about her Journey toward Wellness and will be back with another article in the next newsletter.]

Cynthia Gracey is circumspect about her journey toward wellness. “I have often wondered what I would have done if my first serious attempt to address my symptoms of Charcot-Marie-Tooth syndrome with an alternative therapy had NOT worked” Gracey muses. Charcot-Marie-Tooth (CMT), a hereditary, degenerative neuromuscular disease, had made her legs weak, walking difficult, and twisted her back and pelvis. Gracey had rejected doctors’ recommendations for surgery, striving to find other means to improve her health.

Her first foray into what she calls the “almost mystical land” of holistic healing was a fantastic success. She tried the Roling method (deep muscle massage), which stabilized her body, and she has never had to look back.

Her next great therapeutic bodywork experience was through a nurse at the Muscular Dys-



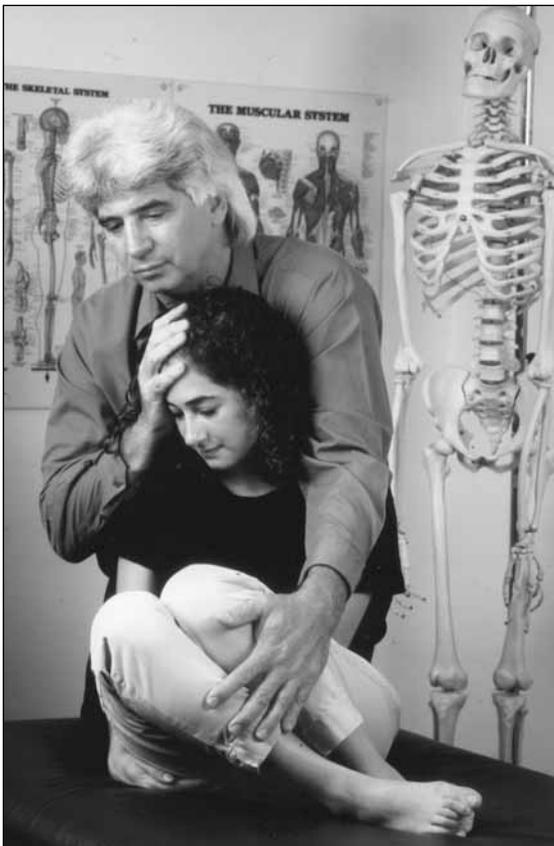
Left: CMTA Board member, Cynthia Gracey poses with the values compass that the Board of Directors of the organization helped create at their retreat in February. The circle represents the values and vision of the CMTA with hope being the core value and the basis of the CMTA’s vision being to generate HOPE.

trophy Society, who was studying a new form of therapy—CranioSacral Therapy¹—with its developer, Dr. John E. Upledger. Having become a believer that the next step in her healing process would be offered clearly and usually in an unexpected way, Gracey decided to try it.

“While the Roling method helped on a very physical level, CranioSacral Therapy felt like it was going deeper, almost into my emotional and spiritual neurology,” she says. “I found it to be like a fine-tuning, encouraging my body, mind and spirit to embrace this body of mine, and to wake up and do the healing work that was necessary. I experienced an expanded sense of well-being, vitality and joy after a session of CranioSacral Therapy.”

Gracey incorporates other bodywork methods, such as hydrotherapy and neuromuscular therapy into her wellness routine along with weight training, breath work, meditation, imagery and nutrition. A former attorney, she now devotes her time to her company, PATH Consulting, and her family—pursuits that complement her healing philosophy.

“Grace, knowledge, intuition and the need to just do that something guided me,” Gracey says, and she busily supports others in their health quest from her home in Florida. “My healing journey, through both alternative and traditional terrain, continues to this day.”



Lower Left: Chas Perry, PhD, demonstrates a technique of CranioSacral Therapy on a patient at the Upledger Institute, Inc. in Palm Beach Gardens, Florida.

¹ The CranioSacral system consists of the brain, spinal cord and dural membrane that surrounds them. Cerebrospinal fluid which nourishes the central nervous system is produced and then reabsorbed in the brain. The production and reabsorption of this fluid produces the CranioSacral rhythm which can be felt anywhere on the body. Cranial Therapy uses this rhythm as a guide to find restrictions in this system and in the connective tissue throughout the body, then uses gentle pressure to free these restrictions.

CMTA Support Group News

Right: Members of Robert Buddé's support group in Lexington, KY, gathered to listen to a presentation on maintaining function with CMT. The physical and occupational therapist discussed an exercise regimen and answered questions from the audience.

Below: Members of the support group from the Church of the Brethren in Greenville, Ohio. From left to right, they are Peg Willis, Dot Cain, Delores Houston and Joe Whitacre. All four of these members wear AFOs.

■ Arkansas - Northwest Area/Springdale

Libby Bond and her support group operated a display booth at the Bella Vista, Arkansas, Health Fair on April 26th and 27th. They had brochures from the CMTA and pens with our logo to give out to visitors to the fair.

■ California - Berkeley Area

Ruth Levitan reports that the last meeting had 18 attendees discussing the names of some good area doctors and the sharing of ideas for coping with CMT. The "spunky" award went to Kim Snadow for coming to the meeting in what Ruth described as an "awesome" brace on her knee and leg. She and her mother, Julia, are dealing with her dislocated knee. A call to the office from a member of Ruth's group resulted in the following compliment: "Ruth has been like a guardian angel to me in the last year. I don't know how I would have coped without her. She's really on a campaign to make doctors here in California recognize and respond to patients with CMT." Way to go, Ruth!

■ Florida - Miami/Ft. Lauderdale

Al and Marilyn Kent hosted the Teddy Bear Auction on March 14, 1999, and although attendance was much less than hoped for, they contributed a total of \$1200 to the CMTA from the proceeds of the auction.

■ Kentucky/Southern Indiana/Southern Ohio

Robert Buddé's support group also had 18 people in attendance, 12 of whom are CMT patients. His group brags that one of their newest members, Howard Beers, is 94 and may well be the oldest CMT patient. (Any challenges to that claim?)



The speaker was Sharon Couch, a physical and occupational therapist at the University of Kentucky clinic in Lexington. She gave a comprehensive talk on maintaining function with CMT. She handed out an exercise regimen and answered questions after her presentation.

The next meeting will be on May 15, 1999, at the Lexington Library, Northside Branch, where the guest speaker will discuss orthotic devices and other equipment to help people with CMT.

■ Michigan - Flint

Debbie Newberger and Brenda Kehoe report that most of the calls they get are from families with small children with CMT who live at least an hour away from them. With the unpredictable winter weather in Michigan, they have decided to meet spring and fall only. Places and dates of meetings will vary depending on the schedules and locations of people who call to attend. If you live within driving distance of White Lake or Mt. Morris, MI, call Debbie at 248-666-9207 or Brenda at 810-687-3368 for details.

■ Ohio - Greenville

Joe and Donna Whitacre, who are members of the group that meets at the Church of the Brethren in Greenville, sent along the picture of four of the members of the group, all of whom wear AFOs. The Whitacres found the support group from the listing in the newsletter and report that belonging to it has been "great!" The Whitacres sent along recipes for the cookbook project as have many other support group members.

■ New Groups...forming and NEEDED!

Several people have received the packet on "how to start a support group" and we are hopeful that we will be able to report first meetings of many of these groups in our next newsletter. Still desperately needed are groups in Philadelphia, New York City and the Los Angeles area. If you live in one of those locations and can commit to getting a group started, call the office at 1-800-606-2682.



CMTA Support Groups

Alabama/Greater Tennessee Valley

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

Arkansas—Northwest Area/ Springdale

Place: Harvey and Bernice Jones
Center for Families,
Springdale
Meeting: 3rd Saturday of each month
Contact: Libby Bond, 501-795-2318

California—Berkeley Area

Place: West Berkeley Library
Meeting: Quarterly
Contact: Ruth Levitan, 510-524-3506

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

California—Napa Valley

Place: Sierra Vista Convalescent
Hospital, Napa
Meeting: Quarterly
Contact: Betty Russell, 707-747-4864

Florida—Boca Raton to Melbourne

Place: Columbia Medical Center,
Port St. Lucie
Meeting: Quarterly
Contact: Walter Sawyer,
561-336-8624

Florida—Miami/Ft. Lauderdale

Place: North Broward Medical
Center, Pompano Beach, FL
Contact: Al Kent,
954-742-5200 (daytime) or
954-472-3313 (evenings)

Kentucky/Southern Indiana/ Southern Ohio

Place: First United Methodist
Church, Lexington, KY
Meeting: Quarterly
Contact: Robert Budde, 606-255-7471

Massachusetts—Boston Area

Place: Lahey-Hitchcock Clinic,
Burlington, MA
Meeting: Every other month, the first
Tuesday
Contact: David Prince, 978-667-9008

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 each month
except January, July, and
September
Contact: Ardith Fetterolf, 816-965-0017,
fax: 816-965-9359

Missouri—St. Louis Area

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

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

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

Place: Brooks Assembly of
God Church
Meeting: Monthly
Contact: Regina Porter
503-591-9412

Texas—Dallas/Ft. Worth

Place: Harris Methodist HEB
Hospital
Contact: Greta Lindsey
(817-281-5190) or Shari
Clark (817-543-2068)

West Virginia/North Central

Place: VFW Conference Room,
Elkins, WV
Meeting: Quarterly
Contact: Joan Plant, 304-636-7152
(evenings)

CALL FOR PARTICIPANTS

I want to thank all of you who responded to the recent CMT and Pregnancy Questionnaire in the December 1998 *CMTA Report*. While many responded, I have not yet received enough surveys for meaningful analysis. Again, I am interested in both those who noted weakening during pregnancy and those of you who did not. If you still have the issue of the newsletter with the survey, please fill out the questionnaire on pages 5 and 6. If you did not get that newsletter, please call the CMTA at 1-800-606-2682 to request a copy. I would appreciate your completing the survey and sending it to me.

Thanks to all of you who have already completed and returned the questionnaire.

Ted M. Burns, M.D.
University of Virginia
Health Sciences Center
Room 2735
Charlottesville, VA 22908





Comments from the CMTA Ho

Editor's note: For those of you who do not regularly (or ever) log on to the CMTA home page at www.charcot-marie-tooth.org there is a bulletin board where interested persons post questions and get answers from others who've been through the experience or have some personal knowledge on that topic. The people who answer are almost never medical professionals, but the answers are amazingly accurate and helpful. We present a few here to give you a taste of what you might be missing by not using the "bulletin board." For first time visitors, go to the home page, address above, and choose "bulletin board" from the column on the left. Once in the bulletin board area, you can post a question or look at any previously posted questions and answers by highlighting the question from the list at the top and then double clicking to open that question.

ANKLE FOOT ORTHOSES (BRACES)

Question: *My husband was diagnosed with CMT about 14 years ago and now has pretty severe leg weakness and severe foot drop. He has never chosen to try AFOs, but recently went to the hospital with severe trauma to his left arm from a fall, probably precipitated by his CMT. While there, the physical therapy people watched him walk and molded an AFO for him. His left arm is in a cast and he cannot move it or even put any weight on it for 6-8 weeks. He is supposed to be starting physical therapy to learn to walk with the right leg AFO and is now finding that he virtually has to learn to walk all over again. The effort this takes is huge and he is fearful of falling on the left arm. I am wondering what the normal time would be to learn to use AFOs. How do people like them? Any opinions on whether it would be better for him to push to learn walking with the AFOs now or when he is healed?*

Response: I'm sorry to hear about your husband's fall. I've had some physical therapy recently and used parallel bars. Perhaps your husband would feel safer if he learned to use his AFO with the bars. My own experience includes AFOs for almost two years. I'm a nurse and I believe my AFOs have allowed me to keep working. Mine are heavier than most molded plastic with built-up ankles. The ankles are hinged so my gait is more normal. My only problem is that I haven't figured out a way to hide them when I'm wearing dresses. I started wearing my AFOs four to six hours a day after several days of checking for red spots that might lead to blisters. Next, I used them for 13 hours a day so I could work a twelve hour shift. Sometimes, my ankles felt crampy, but I had no other problems. My fatigue level was greatly improved and my balance was much better. I have never fallen with my AFOs on. Once, without them, I fell and hurt my foot so badly that it swelled up and two toes turned purple. After that initial injury, my foot only swelled when I didn't wear the braces. Do whatever works best for your husband, but don't give up on a properly fitting AFO which may allow him to be independent longer and prevent future falls.

Response: Like everyone else has said, AFO experiences are different for everyone, but I must agree that trying to adjust to both a broken arm and AFOs at the same time can be very cumbersome. Still, I highly recommend the AFOs! They have also saved me from falling

You can reach the CMTA home page at www.charcot-marie-tooth.org

many times. Fortunately, what falls I have had have not resulted in any broken bones. I am on my second set of AFOs and the first were nearly impossible to adjust to. Luckily, I was able to locate an orthotist who is an expert in the specific needs of a CMT patient. He recommended I start wearing them only an hour a day until I was comfortable with that hour. Then, increase the time by 1/2 hour until I reached a full day of use. I'm only up to 3 1/2 hours daily.

Many general orthotists do not realize just how tender the bottom of our feet are, and therefore, do not make concessions for that in our AFOs. My orthotist put a foam padding in the foot and upper ankle of mine for extra softness and cushioning. I can walk fine now without my cane when I have on the braces. I also use a wheelchair if I'm going to be walking or standing a lot. Unfortunately, it will probably be trial and error for your husband until he gets comfortable with the "new" situation.

Response: In 1991, I fell and broke my left elbow very badly, so I can empathize with what your husband is going through.

About the AFOs, I have worn AFOs since 1975 and I did not have much trouble adjusting to them. At various times when I have had to have an AFO replaced because they broke, there have been several trips back and forth to the orthotist for adjustments. But I would not trade my AFOs for anything. They make all the difference in the world in how I can walk, and, therefore, my self-confidence.

For the last 12 years, I have also used a crutch to aid in walking. A crutch works best for me because my hands are weak and the support under my arm is important. Perhaps your husband could try a crutch or cane on the right side for support and balance, at least while the left arm is healing. I can certainly understand his fears of falling on his left arm!

I think that people who don't have CMT or who aren't familiar with it do not realize how much those of us who do have it depend on our arms and hands to balance ourselves by holding onto rails, walls, doors, etc. Therefore, to have an arm in a cast or sling or otherwise restricted can be terrifying for people with CMT. You might want to point this out to the doctor or therapist.

me Page Bulletin Board

It takes a lot of energy, emotionally and physically, to attempt either of the things your husband is facing...recovering from a bad injury or adjusting to AFOs. It is probably very difficult to try to do both of these at the same time. It might be best to postpone the AFOs, but I would definitely want him to give them a try. They could transform his life!

"HOW DO YOU TELL...?"

Question: *This is the first time I've accessed this forum and I've learned a lot from your previous posts. There is one subject I haven't seen covered though, and that's how do you tell people you have CMT? I've had it since birth, but although my feet and lower legs are pretty badly deformed, with trousers and the right shoes (or more usually, boots) I can fairly well cover it up. I walk quite badly now, and I find it very hard to keep up with other people, as well as not wanting to walk long distances because of pain, but I don't think most people realize there's anything seriously wrong. That helps in some ways, but it means that I have to avoid any kind of situation where I'd have to show my feet, because I can just imagine people's embarrassment if I suddenly revealed them! It also means that because people don't know what's wrong, they tend to think I'm just rather lazy and can make quite hurtful remarks. I play along with that idea because it seems less embarrassing than announcing the truth, but obviously I'd prefer if I could just tell people and, hopefully, get a little understanding. Trouble is, because it's not immediately obvious, there never seems to be a good time to explain—you can hardly announce it, within the first five minutes of meeting someone, but then if you don't say something early on, when do you say it? For example, I've worked at the same office for ten years, and none of my colleagues knows I have CMT (though I guess many of them must know something's not right in the leg department) and I'd feel weird telling them now. I know in many ways this must seem like a non-problem compared to some of the things we go through, but I'd really appreciate any tips.*

Response: This is a great question. I tried to explain CMT to a supervisor when I was first diagnosed last year. The supervisor was also my friend and I REALLY wanted to tell someone. But, I now regret offering the information. My regret is that I feel as if I am treated differently, and although it is not strongly negative in tone, I want to be viewed as the same as all the oth-

ers. I suggest that you offer only information as requested by co-workers. But, I also suggest to start with "I have a condition that affects the strength and motor skills of my hands and feet." Then, if the person is more interested, they will ask questions. I think that saying it is "neurological" is more information than necessary and can really create confusion if the dialogue does not flesh out all the subtle details. Of course, this will vary with the severity of the CMT progression, primarily in how visible it is to others. For those of us that can hide the appearance of CMT, we should be very thankful the progression allows us a "normal" lifestyle. In the lottery of disease in this world, my CMT is so "minor" and I thank the man beyond the clouds for allowing me a "normal" appearance. Good luck. I draw a lot of mental strength from this forum.

Response: I just tell people that I have an hereditary neurological disorder and it causes me problems in walking distances. But, I don't announce it "out of the blue", usually something will precede my telling someone; for example, someone may want me to walk a longer distance than I can and I say that I can't walk far easily because of a neurological disease which affects my feet and legs.

Adjust your comments to necessary information for the individual you are telling. Some people only want to know why you can't walk as far or as fast as they do. Some people will ask more questions. If they do, tell them as much as you think they want to know or you want them to know. I've had the misfortune of falling at work, (I tripped over my feet) and then an accident report has to be filled out. I explained to my supervisor about CMT and the way it makes it more difficult for me to walk. So, take advantage of small opportunities to let people know when necessary.

Response: I basically agree with the other replies about telling other people about CMT. I don't volunteer information unless someone asks. At that time, I will respond with a general description—then if they question further, I will go into more detail. When children ask me why I walk the way I do or why I use a crutch, I tell them I was born this way, but that it doesn't hurt (which for me is true). I have found that if children know it doesn't hurt, they usually don't ask further. I just try not to make a big deal of my handicap, but I will tell others if I encounter steps without rails that I need help with.



Members of the CMTA's Medical Advisory Board answer questions from readers.

Ask the Doctor

Dear Doctor,

Does anyone have any suggestions for dealing with hand pain? As a graduate student, I do a lot of typing at this time of year and my hands are sore and swollen. I take Advil every 4 hours and ice my hands at night, but they still bother me.

The Doctor replies:

Anti-inflammatories and non-steroidal analgesics are appropriate, but should be taken with due attention to the instructions. Better, would be to ask an occupational therapist who specializes in upper body/arm problems for advice about posture. For instance, your keyboard may be at the incorrect level.

Dear Doctor,

I have been suffering with CMT 1 for the last five years. My hands and feet feel like pins are being stuck in the nails. I have fallen 52 times, requiring sutures to many of the cuts on my legs. I have been in 5 hospitals and have been seen by 23 doctors. After much persistence, I was prescribed Tegretol, 200 mg three times a day. This has stopped the "jumping" in my arms and legs. I still have numbness, however.

One neurologist said I was taking far too many medications, but did not advise my family physician what to eliminate. The following are the medications I am taking:

Zoloft 50 mg
 Xanax .5 mg
 1 in the am; 1/2 at supper; and 1 at bedtime)
 Synthroid .5 mg
 Prednisone 10 mg
 Elavil 100 mg at bedtime
 B12 vitamin, 1 per day

I would appreciate your opinion concerning these medications. The Zoloft does concern me. I have been taking it for five years as well as the Prednisone, which has made hair grow on my face. I have lost so much weight that my scalp actually protrudes through my forehead. Physical therapy has helped very much, but even that hurts so badly that I wonder if it is worthwhile.

A Neurologist replies:

Regarding your medication, the Tegretol is for pain relief and is a correct dosage but needs monitoring. It is important to know your age to fully evaluate expected medication effects, but using the Zoloft (a cautioned drug for CMT) may also be causing the extreme weight loss.

You should consult your psychiatrist about taking a different tranquilizer-antidepressant. We will send a list of proscribed drugs for CMT, but maybe your doctors should have copies of the CMT manual for physicians.

The prednisone is not a high dose, but you did not indicate why it was necessary to take it. If it is essential, perhaps it can be given on alternate days. Only one doctor should prescribe medications. You are taking three antidepressants! The neurologist's comments are probably correct.

Dear Doctor,

I stand on my feet most of my 40-hour work week. At the end of the day my legs and feet are very tired and I know the value of swimming as exercise, but am a little reluctant to go to a pool because of the danger of falling when I have to take my braces off and walk to the pool's edge. It is also difficult to get to a pool each day. My question is this: Would there be any value to using a jacuzzi or hot tub each day? I know I have felt better after being in warm water and I wonder if there is any evidence that warm water is therapeutic for someone with CMT.

A member of the Medical Advisory Board replies:

There have been no controlled studies of heat and CMT, but many patients are helped by warm water and gentle movement, similar to athletes. Rough jacuzzi movement may not help and damage can occur with very hot liquids, as numbness may prevent recognition of heat. If you use a pool, you should leave your braces and a towel nearby. Finally, a visit to a rehabilitation doctor (physiatrist) could result in a prescription for hydrotherapy.

Dear Doctor,

My daughter, who is 13 years old and has CMT I, was diagnosed with hip dysplasia last spring. We had corrective surgery done in May at Johns Hopkins. It is now January and she has not healed. Two months ago, it seemed like she was healing and she went to full weight-bearing. This has not happened. She seems to be doing somewhat better physically, but it shows on the x-rays as little or no improvement.

We are sending her to the PT in hopes, again, of stimulating healing. I am also trying to get as much calcium and other nutrients in her. The doctor has said that the next step would be a sure thing. Any opinions would be helpful. The thought of her going

Carnitine and coQ10

back into a body cast again is awful. Does the physical therapy seem like a good idea?

Should we try and wait this out as long as possible before trying a bone graft? If we go with a bone graft, should we go cadaver or use her own?

An Orthopaedic Surgeon replies:

The main problem is whether there has been good communication between the father and the treating physician. Hip dysplasia, although rare in CMT patients, has been seen and in his daughter is a problem. Since I do not have the background information, it is very difficult to know why, just at this time, age 13, she had corrective surgery and exactly what was done. Generally, there should be some sort of reconstruction and that does usually require a bone graft. Nevertheless, it seems that the surgery did not work out fully, and there is a healing problem, which, again, from the note, does not indicate what it is - pain, limping, instability, loss of correction?

There are different ways to try to stimulate further healing, if that is the problem. As a follow-up and, if this is the main concern, it is important to know what has been prescribed, rather than what the child is getting. CMT children do not, generally, need nutritional supplements.

If further surgery is suggested, it would be important for the father to get the following questions answered to his satisfaction:

1. Exactly what is to be done—type of surgery, bone grafting from where to where, etc.
2. What are the problems that might occur if surgery is performed?
3. What is necessary postoperatively—cast, PT, bone stimulation, etc.
4. What is the recovery time period and at what stages could we expect, for instance, removal of cast permanently, crutch walking, weight bearing, etc.
5. What are the alternatives to surgery?
6. What might happen if surgery is not done?
7. What complications might be expected if surgery is not done?

All these questions constitute an “informed consent” and, in California, this is what we must disclose to our patients before we operate and put in their records.

*John Hsu, MD, Orthopaedic Surgeon,
Rancho Los Amigos*

An article in *Quest*, the publication of the Muscular Dystrophy Association, discussed these two popular supplements in an article entitled, “Miracle Cures or Money Down the Drain.” We present a brief summary of some of their findings:

Advertisers and some researchers claim that carnitine and coQ10 can increase muscle strength, treat a number of heart conditions, increase overall energy levels and add to the body’s ability to fight free radicals.

Just because these compounds are “natural” and involved in certain bodily functions doesn’t mean that taking more will improve normal activities, the author points out. Sometimes the body is only capable of absorbing so much of a substance and no more, in which case, as one doctor pointed out, you’ll just produce “expensive urine.”

Both doctors interviewed in the article stress that carnitine is only helpful for someone with carnitine deficiency, or when medication or diet results in a deficiency. When it is used for general tiredness, fatigue, improved athletic performance or ailments without a deficiency, there is no evidence that it is justified or useful.

CoQ10 is made in all the cells of the body. Scientists believe that coQ10 works as an antioxidant in the cells and neutralizes the charged oxygen cells without becoming destabilized itself, thus stopping the destructive process of oxidation. The therapeutic benefits of coQ10 (cited by some as preventing heart disease, cancer, neurodegenerative diseases and old age) remain unknown because of a lack of controlled studies. The good news is that coQ10 has no known side-effects and it is available in health food stores and pharmacies. The daily dosage of 30 milligrams should cost about 33 cents a tablet.

Editor’s note: Just days after the above information was published by the MDA, USA Today, (March 10, 1999), published an article on creatine and its potential to strengthen muscles in the elderly and those afflicted with diseases such as muscular dystrophy and ALS (amyotrophic lateral sclerosis). Excerpts from that article follow:

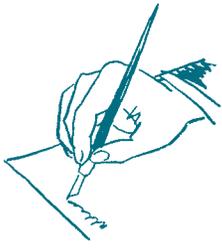
Creatine, the controversial dietary supplement popular with muscle-building athletes, has been found to help patients with diseases such as muscular dystrophy and ALS, Lou Gehrig’s disease. A Canadian study found that these patients had a 10 to 15% improvement in their ability to perform high-intensity exercises while taking creatine. The creatine does not arrest or reverse the disease process, but gives the muscles more energy.

Questions about the safety of creatine continue, especially for athletes taking large doses. There is some concern that it could harm the kidneys and other organs. The Food and Drug Administration says long-term effects of creatine are not known. The FDA recommends that consumers consult with their physicians before taking this or any product like this.

The study was conducted at McMaster University Medical Center in Ontario, Canada. Researchers gave 5 to 10 grams of creatine for ten days to 81 people with diseases that cause muscle weakness and atrophy. Their strength went up on every measurement. There are implications that this treatment might also help the elderly who are weak and in danger of falling and breaking their hips.

Note: This study did not involve patients with CMT, so there are no known results for people with CMT. As with all treatments, you should consult your doctor before beginning any new program.





WRITE TO US!

Pat Dreibelbis, Editor

The CMTA Report

CMTA

601 Upland Ave.

Upland, PA 19015

*The CMTA reserves
the right to edit
letters for space.*

Letters to the Editor:

Dear CMTA,

Some years ago, my brace maker and I designed a pair of braces that I could wear in the shower and which would also be easy to put on if I had to get up at night. In general terms, they are AFOs with rubber soles and a piece of plastic over the instep. A Velcro strap at the top and across the instep hold each brace secure enough for wearing around the house.

In recent years, the erosion of my gait, balance, and hand strength has made it clear that if I am to continue to travel, I must lighten my luggage. Bringing that second pair of braces with me means I have to use a large bag to accommodate them and a second bag to carry all the things they displace in my big bag. I have no intention of letting CMT stop me from traveling, yet. I'll be 70 this year.

How do other people deal with the problem of getting in and out of the shower without feeling as though their regular AFOs turn into ice skates when they come into contact with a hard or wet surface? I broke my leg going into the shower without my AFOs, so that's what motivated me to figure out something so I could wear braces in the shower. I'll bet there are some pretty ingenious solutions out there! What I'm looking for is a way to adapt my AFOs so I won't have to carry the second pair.

I've spoken with my brace maker about putting a piece of plastic across the instep of a pair of rubber-soled sandals with Velcro to fasten the instep and at the heel. I would be able to slip these onto my AFOs and be secure in the shower and on hard surfaces. These "slippers" would be less bulky than a second pair of AFOs, but I would like to hear what others have figured out.

—B.D. CA

Dear CMTA,

I do appreciate receiving *The CMTA Report*. It has helped me a lot to understand what has, and still is, happening to me.

The doctors in West Virginia have little knowledge of CMT. I have made copies of articles and given them to several doctors. I was diagnosed in Charleston, WV, in 1990. In February, 1991, I lost my good leg (the left) to diabetes. I have a prosthetic made by Nova Care. The expenses are terrific! My main goal in life is to keep my right foot. Special shoes and an orthotic for the precious right foot cost a lot. I receive no help from Medicare or insurance for the above items. My right shoe is rebuilt to fit my

crooked, crippled foot by a pedorthist in Pittsburgh, PA.

I really appreciated the letter in the December *CMTA Report* from "D.W.—CMT challenged and incredibly stubborn." How very true! I'm too stubborn to toss in the towel. I just get mad and try harder. I agree that the mule would be a great mascot for us.

Please keep up your work. There are a lot of us out here.

—M.L.S. WV

From the Internet:

Hello everyone. I had a bad experience after school today. I was walking out and two boys started making fun of the way I walk. I usually just ignore this, but I just had to say something today. I am very sick and tired of this happening to me and other people. I had a few words to say to them (not very kind) and then I told my Mom. We went to the secretary and she said the boys would see the principal. I also have had a bad experience at the mall. It makes me sick to think and see how cruel the society we live in today, is. I hope that someday people will realize we can't help how we walk or look in any way, shape or form and look for inner beauty instead of outer.

—K.R. (14 years old)

A 10-year old replied to her:

No one ever teases me, but when I go to P.E./gym, I feel awkward because I can't run fast. Other kids used to ask why I run funny, but my coach talked to them. Now, I only have to do half of what everyone else does. If the other kids still tease you, tell your teachers and your principal.

Dear CMTA,

I wish to raise the issue of Long Term Care Insurance for those who have CMT. My own experience is revealing and probably not atypical.

I was diagnosed with CMT at age 50 (now 70). The usual symptoms surfaced about age 45. Since then I have progressed to AFOs and a cane when I anticipate I will have to stand in one spot. Other than that, I am completely self-reliant and functional. About two years ago, I applied for a LTC policy with my pension holder. I acknowledged on my application that I had CMT, and, of course, that was further documented by my medical records. I was visited by a social worker who gave me several verbal tests and for whom I demonstrated by walking that I was quite functional. Six months after my appli-

cation, I received a note of rejection based on my CMT. I spoke with the underwriter at the insurance agency, who informed me that he had automatically rejected about six previous applicants for the same reason. I appealed his decision and he, after a dialogue with his medical consultant, confirmed the rejection on the phone.

As you and I know, the degree of disability among those with CMT varies greatly and a diagnosis of CMT does not a priori indicate a need for living assistance. Most physicians are very ignorant about CMT and I imagine that applies to the medical consultants advising the insurance industry. What can be done to inform them and thus enable them to make a more informed risk analysis of individual cases?

In addition to sponsoring research, an important function of the CMTA is providing a forum for information of interest to our community. You are doing good work.

—R.L. CA

Editor's note: Do any readers of the newsletter work in the insurance industry and have insight into what the organization might do to educate these medical consultants? We have tried to work in the area of health insurance in the past and have found the insurance companies vary so tremendously from state to state that any kind of uniform approach was impossible. We'd appreciate any insight our readers might provide.

Dear CMTA,

I have CMT and the following problems. I'm wondering if any other CMT patients experience any of these conditions? The right side of my heart is larger than the left side. I have also lost all sense of smell and when I shower, I have so little sense of feeling on the back of my neck that hot water feels cold when it is not.

—T.G. AZ

Editor's note: This question was posed to a member of our Medical Advisory Board and his response was that a small percentage of CMT patients have EKG conduction abnormalities but right heart enlargement would suggest chronic lung disease such as emphysema. The other symptoms are not consistent with CMT and an upper cervical spine or brain stem disorder should be evaluated.

Dear CMTA,

I am 25 years old and have lived with CMT all of my life, although I was not diagnosed until the age of 20. Since then, I have gone through a series of seven surgeries on both feet to correct

the deformities common in CMT. I'm writing because where I live (Jamestown, NY), there are no support groups of any kind. I'm looking for anyone from my area (or elsewhere) that I could correspond with between the ages of 18-30. If anyone knows of a chat line for CMT on the Internet, please let me know.

Write to Raymond Bigney, Jr. in care of the CMTA office at 601 Upland Ave. Upland, PA 19015.

Dear CMTA,

I'm sure you have heard many stories from people with CMT. I found Cynthia Gracey's article in the last newsletter very interesting and hopefully, many CMT patients have had the fortune of reading it. I am deteriorating faster than the "uneducated" doctors believe and I have been told that this is not normal. I am very in tune with my own body. I have always had a balance problem and looked for something to grasp on to so I could keep from falling. I've taken my share of tumbles and falls.

It has taken me 1 1/2 years going from doctor to doctor looking for pain relief, but have now found one who keeps my pain at a moderate level so I can be semi-functional in life. Most of all, I thank God that He is there for me. If anyone with CMT would like to write to me, I can be contacted at Debbie, PO Box 245, Granite Falls, WA 98252.

Dear CMTA,

I could recite a litany of wrong diagnoses, dire predications (including death at 35) and poor remedies.

What I have to offer is my own present solution for functioning with CMT.

I purchase a shoe called a chukka boot from Hitchcock Shoes, Inc. Hingham, MA. It is deep enough and wide enough to accommodate an orthotic. I am 6' tall, but my shoe size is 6 1/2 x 6E. I take the shoe to the orthotist at Wright & Filippis, in Rochester, MI. Mr. Unger designs an orthotic and modifies the sole of the shoe to give me more outside support. I also have a Jazzy electric wheelchair made by Pride Co. in Exeter, PA. This chair has a lift seat that raises me high enough to stand erect. Once on my feet, I can use my walker.

At age 82, if I can help anyone avoid the dead ends that I have encountered for more than 50 years, this letter will be worth writing.

—J.C. Bloomfield Heights, MI

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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, Macrochantin)
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, 1X, and HNPP 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*



601 Upland Avenue
Upland, PA 19015
1-800-606-CMTA

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