

The CMTA Report

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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South Florida CMT Conference Co-hosted by The University of Miami and the CMTA

On Saturday, February 7, 1998, 128 patients, family members, students, and medical professionals attended the conference on Charcot-Marie-Tooth disorders at the North Broward Medical Center in Pompano Beach, Florida. The day began with an overview of the history of Charcot-Marie-Tooth disorders and the current status of research and knowledge about the disorder from Dr. Walter Bradley, neurologist, from the University of Miami. His presentation was followed by a panel of experts: Dr. Lisa Baumbach-Reardon, University of Miami, who discussed CMT Type I, Dr. Jeffrey Vance, Duke University, who discussed CMT Type 2, and Dr. Rudolph Warner, University of Miami, who concluded with information on CMT X-linked. All of the presentations were followed with question and answer periods.

Following a morning coffee break, the large group divided into smaller groups for the workshop presentations. Topics were pain management, alternative therapies, orthotics and pedorthics (bracing and shoe-fitting), and physical and occupational therapy. Interest in the workshops was very high and the group in the alternative therapy session voted to delay lunch so they could stay longer and learn more. The CMTA wishes to thank Frank Crohn for funding the conference meals.

After lunch, Dr. William Quinn discussed orthopaedic surgery and non-surgical options for treating deformities of the foot and leg. He was followed by Dr. Richard Rogachefsky, a hand surgeon, who discussed carpal tunnel surgeries and tendon transfer surgeries to alleviate wrist and hand pain as well as correcting finger/thumb pinch. Amy Kalick, Athena Diagnostics, concluded the afternoon group pre-



Sisters Leonor Deetjen and Marie Fernandez de Velazquez "won" the award for traveling the greatest distances to attend the Florida conference. Leonor journeyed across the country from California, only to be outdistanced by her sister, Maria, who came from Spain to attend the conference.

sentations with a discussion of the diagnostic DNA testing available through Athena's labs.

The group then broke again into the small workshop settings, where they could choose another topic from the four offered in the morning. The day concluded with closing remarks by Dr. Robert Lovelace, Chairman of the CMTA's Medical Advisory Board.

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Begin With Flynn

By **PAUL R. FLYNN**, Executive Director

Exec. Director's Note: At the risk of seeming immodest, I chose the above heading for both the placement of my article at the beginning of the newsletter and the fact that it is short but catchy...or so I think. Hopefully, you will forgive this indulgence.

*They say, to know a man,
you must walk a mile in his shoes.*

Walking in the shoes of an individual with CMT would be a start toward understanding his or her life, particularly if the shoes were custom-made or worn with some type of orthotic device, but that would hardly provide a true picture. As I conclude six months on the job as the first executive director of the CMTA, I realize that I have much to learn. While I have CMT, and come from a large family affected by the disorder, I know that this life experience is still limited. Many of you have provided me with valuable insight into the highly unique and personal challenges of CMT as it relates to yourself or a loved one. The anecdotes you provide me are a tremendous source of "relevant data" in a practical sense and great inspiration in a spiritual way. I encourage you to share your experiences with others through our newsletter, the Internet, or with me directly, as it will advance all of our understanding about our misunderstood condition.

The Dreamcoat Singers will perform in a benefit for the CMTA at the Westchester Broadway Theater on Tuesday, March 31, 1998. Please contact the CMTA office for ticket information or to join the event committee.



Dr. Paul Donohue: Dr. Donohue has a syndicated column that appears in nearly 300 newspapers around the country. He responds to a wide variety of medical questions in his column, which typically appears in the section of the newspaper with Dear Abby and Ann Landers. Dr. Donohue's article about CMT first appeared in early January and generated an incredible response. Nearly 900 calls flooded the CMTA's 800 line over a two-week period. For those of you who heard of us through Dr. Donohue, welcome to the Association!

Advertising and Endorsements: At the February meeting of the CMTA Board of Directors, a decision was reached to allow paid advertising to appear in the newsletter. An increase in requests for ad space along with the Board's desire to maximize this new revenue stream prompted this change of policy. Paid advertisements will be specifically defined as such and will not compromise the integrity and presentation you have come to expect from *The CMTA Report*. As a corollary to this new policy, the Board re-emphasized its position that the CMTA does not endorse or promote any service or product, but merely seeks to provide useful information.

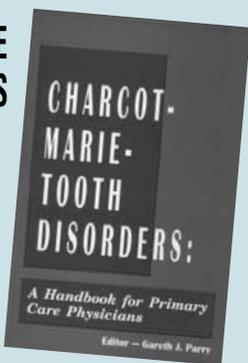
Dr. Frank Palermo and Electrical Stimulation: In the past, *The CMTA Report* has included the findings presented by Dr. Palermo on his electrical stimulation machine as well as letters to the editor from "users" of this machine, both pro and con. Officially, the CMTA is not in a position to validate the efficacy of the electrical stimulation machine. However, in a conversation related to this issue, Medical Advisory Board member, Dr. Stanley Myer, Columbia Presbyterian Medical Center, indicated the risks related to using electrical stimulation on areas with nerve loss, although a risk/benefit ratio is yet to be determined.

Finances and Charitable Support: As a 501(c)(3)-designated charitable organization, the CMTA is mandated to provide information and support to all who request it, regardless of their ability to financially support the organization. While there is a concerted effort to increase the number of supporters and their level of giving, the CMTA recognizes that some members simply do not have the means to give. Recognizing

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Read about:

- *Charcot-Marie-Tooth Disorders in Children*, by Harold Marks, MD, A.I. duPont Institute;
- *The Clinical Features of Charcot-Marie-Tooth Disorders*, by Carlos Garcia, MD, Louisiana State University School of Medicine;
- *Genetics of Charcot-Marie-Tooth Disorders*, by Jeff Vance, PhD, MD, Duke University;
- *Physical Therapy: Conservative Management*, by Carol Oatis, PT, PhD, Beaver College;

and eight more chapters, each written by an expert from the CMTA's Medical Advisory Board.

VIDEO TAPE OF WAYNE STATE CONFERENCE NOW AVAILABLE!

A tape of the Wayne State CMT conference held on October 18, 1997 in Detroit, Michigan is now available. The tape contains over five hours of presentations from the conference. Featured on the tape are Dr. Michael Shy, neurologist and head of the Wayne State CMT clinic, discussing Types I and 2; Dr. William Quinn, podiatrist and member of the CMTA Board of Directors, discussing surgical options; Dr. Steven Hinderer, physiatrist, discussing rehabilitation and CMT; and Dr. John Kamholz, neurologist, presenting an overview of CMT research from an historical perspective. Interviews and excerpts from workshops round out the material. See order form at right.



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

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CMT Facts I <input type="checkbox"/> English <input type="checkbox"/> Spanish		members \$3 non-members \$5	
CMT Facts II <input type="checkbox"/> English <input type="checkbox"/> Spanish		members \$5 non-members \$7	
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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.

BEGIN WITH FLYNN

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this, the goal is to “remind and persuade” those who can give to do so while trying not to offend the rest... a fine line to walk, but we will strive to do it.

The Westchester Dreamcoats: The Dreamcoat Singers are a group of 29 Westchester, New York, children ranging from 9 to 16 years of age. The chorus originally appeared in “Joseph and the Amazing Technicolor Dreamcoat” and performed on Broadway from August 1993 until January 1994 under the name of “The Carolabbe Chorus.” The Dreamcoats will be performing in

a benefit for the CMTA at the Westchester Broadway Theater in Elmsford, NY. The CMTA gratefully acknowledges George Puello from the WBT for contributing his original works, “Scattered Thoughts” and “Parucia” for this benefit event.

Please accept the apologies of the staff of the CMTA for the lack of timeliness in responding to calls during the great “Donohue” flood. The office is still trying to recover from the 900-plus calls and requests for information packets. If you were missed in the chaos that ensued from the national publicity, please forgive us and call again. We DO want to respond to your needs and requests.

Dr. Jose Suarez explains an orthotic device to Mrs. Rani Khanuja while Karen Van Vonno and pedorthist Mr. Ted Mandes look on. One of the great advantages of attending a patient/family conference is the opportunity to speak with noted experts on the treatment of CMT.

FLORIDA CONFERENCE

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On Friday evening, February 6, 1998, approximately 30 of the conference attendees enjoyed a hospitality suite in the Ft. Lauderdale Marriott North sponsored by Athena Diagnostics. The two-hour “get-together” enabled people to get to know each other and to talk one-on-one with the staff and Board members of the CMTA.

Comments on the survey forms that attendees completed suggested that both the opportunity to hear true experts on CMT and to meet and mingle with other people dealing with the disorder rank as the most important aspects of a conference. Plans are now underway for the next

conference, which will be held at Rancho Los Amigos in Downey, California (suburb of Los Angeles) on June 27, 1998. That conference will be hosted by Dr. John Hsu, orthopaedic surgeon and member of the CMTA's Medical Advisory Board.

Persons interested in accessing presenters at the Florida conference may do so by calling the following numbers:

- Dr. Walter Bradley, Neurologist, University of Miami, 1-305-243-7525.
- Dr. Lisa Baumbach-Reardon, Geneticist, University of Miami, 1-305-243-3997.
- Dr. Jeffrey Vance, Neurologist, Geneticist, Duke University, 1-919-681-5696.
- Dr. Rudolph Warner, Biochemist, University of Miami, 1-305-547-6998.
- Dr. Richard Rogachefsky, Orthopaedic Hand Surgeon, University of Miami, 1-305-585-7149.
- Dr. William Quinn, Podiatrist, Plaza Podiatry, Wisconsin, 1-715-341-1266.
- Dr. Robert Lovelace, Neurologist, New York City, 1-212-567-7874.
- Dr. Jose Suarez, Orthotist, Miami, 1-305-595-0567.
- Dr. Russell Bourne, Clinical Psychologist, Upledger Institute, 1-561-622-4334
- Mr. Ted Mandes, Pedorthist, Nobile Shoes, 1-561-842-7400.
- Mrs. Cynthia Gracey, Alternative Therapy, PATH Consulting, 1-561-243-0000.
- Ms. Amy Kalick, Athena Diagnostics, 1-800-537-8899.
- Mrs. Lisa Carlstadt, Occupational Therapist, North Broward Medical Center, 1-954-786-6473.
- Ms. Charlene Henderson, Physical Therapist, North Broward Medical Center, 1-954-786-6473.



CMT Type 2: Report and Update

By **JEFFREY VANCE, Ph.D., M.D.**

In 1968, Drs. Dyck and Lambert separated CMT into two groups, based on neurophysiologic and pathologic grounds. Clinically similar individuals affected with Type 1 have slow nerve conduction velocities (NCV), while Type 2 patients have normal or only slightly slow conduction velocities. Nerve conduction is another term for how fast the nerve impulse travels down the nerve from the spinal cord. This observation reflects a basic pathologic difference between the two types. Type 1 has gene defects that affect myelin, the covering around the nerve axon. Myelin is what controls the speed of peripheral nerve impulses. Thus, defects in myelin lead to the slow nerve conduction speed of Type 1. However, Type 2 affects only the axon, while the myelin is normal. Therefore, Type 2 families have normal or slightly slowed NCV.

As mentioned, Type 1 and 2 are clinically similar. However, there are some minor differences. As a group, Type 2 has a wider range of age of onset and more conserved extremity reflexes. However, in a single individual, a physician cannot tell Type 1 and Type 2 apart based on clinical exam alone.

Genetic studies have greatly contributed to our understanding of CMT Type 1. CMT2, however, has yet to have any gene identified that leads to this disorder. Several reasons have made CMT2 more difficult to study than Type 1. The ability to tell gene carriers using NCV (CMT1 carriers have very slow NCV whether they have symptoms or not) made CMT1 families very powerful for research. However, CMT2 is more difficult to work with, as diagnosis is based on clinical findings alone. It is also less frequent, as CMT2 is thought to represent 1/3 of all CMT cases. CMT2 patients also demonstrate greater variability in symptoms, again making diagnosis more difficult. In addition, unlike CMT1, it appears that no one gene is the primary cause of the disease. This makes fewer families of any one type available for study.

However, in recent years, significant headway has been made in CMT2. In 1993, our laboratory localized the first gene for CMT2 (CMT2A) on chromosome one, at the top of the chromosome (1p36). This led to continued efforts today in our laboratory to identify this gene.

In 1995, Dr. Kwon and colleagues described a family appearing as CMT2, but also with ulcerations and limb amputations as a common factor



in the phenotype. They named this as CMT2B and localized it to chromosome 3. As has been pointed out, this phenotype is different from that of CMT1, CMT2A, and CMT2D, and is thus a rare variant of CMT2. Dr. Peter Dyck and colleagues have reported a third type of CMT2 (CMT2C), which has an additional symptom of vocal cord weakness and respiratory failure. This, too, is a rare form of CMT2, with only a few families reported. In 1997, Dr. Ionasecu and colleagues found a locus on chromosome 7 for a large Iowa family with typical CMT2, and our laboratory has recently confirmed this locus, identifying two additional CMT2D families in our Duke University collection of CMT families. Of 15 large CMT2 families collected at Duke, five families are CMT2A, with two being CMT2D. The rest of these Duke CMT2 families are not localized in any of the four known chromosome regions; thus, additional genes for CMT2 remain to be identified.

In summary, CMT2 has been estimated to comprise about 1/3 of all CMT cases. However, given that it is very similar to many of the idiopathic or common neuropathies seen by neurologists every day, it is likely to be underdiagnosed. CMT2A and 2D are clinically similar to CMT1, while CMT2B and 2C represent rare CMT2 variants. CMT has at least five genes contributing to its cause and likely more.

Given that diagnosing CMT2 is more difficult, the identification of CMT2 genes will greatly aid physicians in CMT2 diagnosis in the future. In addition, as the axon is the final end point between the gene defects leading to CMT1 and CMT2, identification of CMT2 is likely to provide continued insight into the pathology of both forms of this common genetic disease.

Dr. Jeffrey Vance (middle) shares his recent work on CMT Type 2 with fellow Florida conference presenters, Dr. Lisa Baumbach-Reardon and Dr. Walter Bradley.

Summer Research Fellowships:

An Orthotics Survey of CMT Patients

JEFFREY ANDRE BONNEVILLE, MS, University of Health Sciences, Antigua School of Medicine

The goal of this study was to investigate CMT from an orthotic perspective, to increase public awareness of CMT, and to acquire documentation from a survey of patients and orthotists regarding treatment.

Studies exist with regard to genetic pathophysiological evaluations and treatments of CMT. However, to date, the literature does not reflect or attempt to investigate the types of devices used by the CMT population or the efficacy of these devices. Professional (orthotic) impressions have never been evaluated.

We wanted to ascertain what orthoses applications were available and how well they functioned. Perspectives from both the providers and the users needed to be collected. For this reason, we created two separate surveys. From the Certified Orthotists, we requested their criteria for patient applications as well as their professional assessments of orthotic devices already in use. In the second survey, we asked the patients what type of orthotic devices they used, as well as their impressions of effectiveness.

The CMT Patient Survey:

One thousand patient surveys were distributed by the CMTA. Four hundred sixty-two were returned. Of the 1000 surveys distributed to orthotists, 94 were returned.

Of the 462 patient surveys returned, 154 indicated they were diagnosed with CMT Type I, 86 individuals indicated CMT Type II, three individuals indicated CMT Type III, and eight individuals indicated X-linked CMT. Two hundred seven were not sure and four individuals who returned the survey indicated that they had no diagnosis of CMT. However, they were members of the association on behalf of people they knew with CMT.

The median respondent age was 46 years. The average age of diagnosis was 40 years. Two hundred thirty-two respondents were male. With regard to family history, 287 individuals indicated there was a family history, 93 individuals had no knowledge of family history, 75 were not sure, and seven individuals failed to respond to that question.

Predominant Symptoms:

The predominant disabilities indicated in the surveyed patients (many patients had multiple symptoms) were as follows:

- 231 indicated that they had difficulty standing
- 235 experienced numbness (parasthesia)
- 233 believed they had weak ankles
- 206 viewed their disability as severe*
- 124 viewed their disability as moderate*
- 183 viewed their disability as slight*

The table below illustrates the variety of CMT symptoms experienced by the respondents. (See "Orthotics Survey Results.")

The respondents who viewed their level of activity as being extremely limited were 144. Eighty-one individuals stated that they had limited activity and 33 individuals had very few limitations.

Orthotic use was indicated by 232 individuals, while 224 indicated that they do not use orthoses and six individuals left the question blank. Of the respondents who said they used orthoses, the majority used below-knee orthoses, but were unable to correctly identify what type they used. It became clear with the conflicting information amassed by our survey that those CMT patients who were orthotic users, in general, could benefit from orthotic education, with an emphasis on understanding types, designs, functions, and terminology.

ORTHOTICS SURVEY RESULTS

TOTAL RESPONDENTS	SYMPTOMS REPORTED
202	High arches
145	Flat feet
246	Hammer toes
216	Lower extremity involvement (161 indicated right-side weakness)
129	Upper extremity involvement
251	Unable to maintain balance on tiptoes
121	Had no difficulty standing

*The CMTA recognizes that the number of respondents to these questions exceeds the number of participants in the survey. We were unable at the time of publication to clarify the discrepancy.

A Review

The CMTA awarded \$16,000 in summer grant money in 1997. The recipients report, below, on their work. Summer grants are intended to support short-term studies by students who, hopefully, will have their interest in CMT ignited by the summer study and will continue in the field of CMT research or clinical practice.)

Other highlights of the survey:

Two hundred twenty-four individuals believed their devices helped, rather than hindered, and only nine individuals felt that the opposite was true.

Fifty percent of all respondents who answered the open-ended question about orthotics said they did not wear braces and were extremely happy about that. They felt braces were too big, bulky, and cosmetically unattractive. Braces made them feel self-conscious. They would rather walk with an altered gait than walk with a disagreeable, bulky device. Brace wearers used the open-ended question to explain that braces had helped them tremendously, allowing them to continue walking and participating somewhat normally in society. The braces gave many CMT patients a sense of stability and balance.

Only 94 surveys sent to professional orthotists were returned. Low response could be due to hectic business schedules, short notice, lack of interest, or lack of CMT experience. The average number of CMT patients seen by the respondents was four per year.

Conclusions:

Although the responding orthotists were few in number, we felt it was representative. All the orthotists agreed that treatment of the individual was their primary practice, and that there were varied solutions to the common problems of CMT bracing.

Issues noted:

- CMT patients are not always familiar with orthotic terminology and options. Many patients did not use orthotics. Many may not be aware of progressive deformities that may be prevented through aggressive/corrective bracing compared with accommodative devices.
- Because of the wide variation of symptoms in CMT patients, no single orthosis was reported as the overwhelming favorite by orthotists.

However, since the individual is treated and not the disease, the patient's needs must be met first, with an eye on potential progressive weakness.

- There should be some discussion between the educated orthotist and the CMT patient regarding the pros and cons of different types of braces.
- There should be scheduling of regular, follow-up appointments to monitor skin conditions and progressive weakness. Re-evaluation of bracing against such changes could be recommended as well.
- In addition, the perception that a large number of patients had that braces were bulky, and unattractive was not well defined. Did they only have an image of metal orthoses attached to shoes? Had solid ankle-foot orthoses or posterior leaf spring orthoses ever been discussed or demonstrated?

Recommendations:

- Articles should be published to help educate the CMT population on orthotic management and terminology, accompanied by photographs and descriptions of devices presented in a clear, easily understood manner.
- Education, through published articles, in professional journals, and as presentations at regional conferences or seminars can help orthotists recognize CMT as a diagnostic group.

For our research team, this was an excellent learning experience. This study taught us the shortcomings of mixed surveys. Data compilation from limited responses—especially with the orthotist's survey—made us examine more closely the method of our census. Upon reflection, we realize our questions for orthotists would have to be more concise to have a better percent of participation. Conversely, our questions for the CMT patients would, in fact, have to be more detailed for us to receive more specific answers.



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Summer Research Fellowships:

However, we feel we have ventured down a new avenue of awareness. It is our hope that, through our efforts, we may trigger an interest among colleagues and health care practitioners in CMT.

Editor's note: Mr. Bonneville and his associates would like to thank the members of the CMTA who made this study possible by completing and returning the survey they received.

Identifying CMT Families

DR. ELENI ZAMBA, The Cyprus Institute of Neurology and Genetics

The report covers the progress achieved for the support period from 6/97 to 9/97. During that period, I trained and worked at the Department of Neurology, College of Physicians and Surgeons, Columbia University, New York. I mainly worked in the EMG and nerve conduction laboratories under Director Dr. Dale Lange and the Division of Neuromuscular Disease, including the MDA clinic.

I participated in (1) clinical evaluation of CMT families at the MDA clinic, (2) the neurophysiological classification of CMT forms and distinctions between the hereditary neuropathies and the acquired neuropathies including nerve conduction studies, needle electromyography, and report composition, and (3) characterizing the morphological features of nerve biopsies in the pathology meetings at the neuropathology laboratory.

Since my return to Cyprus, I am actively participating in identification of autosomal recessive forms of Charcot-Marie-Tooth and autosomal dominant axonal CMT2 in Eastern Europe and the Mediterranean region under Dr. Lefkos Middleton, member of the CMTA Medical Advisory Board. The following families underwent clinical, neurophysiological and neuropathological evaluation:

1. CMT Type II (autosomal dominant/axonal): one family from Cyprus with five individuals, three of them affected and one family with 10 individuals, seven of them affected, were studied during a field trip.
2. Autosomal recessive CMT: seven families, all of them consanguineous (editor's note: relat-

ed by descent from the same ancestor) from Eastern Mediterranean region were evaluated during a field trip. Thirty-one individuals of these families were affected.

The above families were grouped based on their clinical, neurophysiological and pathological features. Molecular genetic studies are in progress at the Cyprus Institute of Neurology and Genetics.

Classification by Motor Nerve Conduction Velocity (MNCV)

DR. ODILE DUBOURG, Hopital La Salpetriere

Charcot-Marie-Tooth (CMT) disease is a pathologically and genetically heterogeneous group of hereditary motor and sensory neuropathies characterized by slowly progressive weakness and atrophy, primarily in peroneal and distal leg muscles. Two major types have been distinguished, in which the neuropathy is of demyelinating or axonal type. Electrophysiological studies have confirmed the discrimination between demyelinating and axonal forms according to values of the motor nerve conduction velocity in the median nerve (MNCV).

Genes have been identified for CMT1A and B, HNPP (as a deletion on chromosome 17 corresponding to the CMT1A duplication) and CMTX1 (the dominant sex-linked form). All others are loci designations only and include the CMT1C, CMT2A, B and D (four loci), CMTX2, CMT4 (recessive), and some others.

Beginning in 1991, patients from 271 CMT families were examined at La Salpetriere hospital in Paris and classified according to patients' median nerve motor velocities and the mode of inheritance in order to evaluate the frequencies of the different subtypes. The frequencies of the more common mutations, duplication of the 17p11.2 region, PMP22, P0, and Cx32 mutations, were evaluated in the different subtypes. These data led us to determine an accurate threshold value of median MNCV for CMT1, CMTX, and CMT2 and the elaboration of a rational strategy for molecular diagnosis based on median MNCV and mode of inheritance.

A Review (cont.)

In CMT1, at-risk relatives were considered affected if MNCV was less than 30 meters per second (m/s) in one nerve, or between 30 and 40 m/s on two nerves of the upper limbs (both median nerves or the ulnar and median nerves). In the axonal type, (CMT2, etc.) index cases had to present: 1) slowly progressive, bilateral, and symmetrical distal weakness and wasting, in limbs with areflexia (at least of ankle jerks) and distal sensory loss in lower extremities, and 2) MNCV above 30 m/s, with absent or markedly reduced SNAP (sensory nerve action potential). At-risk relatives were considered affected if they had absent or markedly reduced SNAP amplitude in the lower limbs with all or some CMT clinical features. Spinal forms of CMT can be distinguished from axonal CMT by the absence of distal sensory loss and the normality of SNAPs on electrophysiological testing.

The frequencies of the different subtypes were similar to those reported in 1983 by Professor Bouche and colleagues, with a nearly identical proportion of demyelinating and axonal form, 131 and 122 families, respectively. In demyelinating CMT, autosomal dominant and dominant inheritance was observed with similar frequencies. Dominant inheritance was present in 47 axonal CMT families, whereas autosomal dominant inheritance was only observed in 15 families. The predominance of dominant families for axonal subtype is surprising, but could be explained by the fact that it might include numerous CMTX families, in which the histopathological lesion is primarily axonal. In contrast, spinal CMT appears to be rare, as it has been characterized in only six families, five with autosomal dominant, one with dominant inheritance, and seven isolated cases. Isolated cases are frequent for both demyelinating and axonal CMT, 51 and 59, respectively. It is noteworthy that parents were not always examined. Therefore, the term "isolated" should be used, instead of "sporadic."

For all consenting family members, a blood sample was taken for DNA extraction and for lymphocyte immortalization. DNA from an index case in each CMT family was tested for the presence of the duplication of the 17p11.2 region. For patients without 17p11.2 duplication, Cx32 and P0 genes were explored using a rapid non-radioactive technique to detect mutations.

The 17p11.2 duplication was identified in 81 of 123 (66%) demyelinating CMT families tested,

36 of 41 (88%) autosomal dominant families, 30 of 35 (86%) dominant families, 0 of 3 recessive families, and 16 of 44 (36%) isolated cases.

In contrast, this study underlines the very low frequency of the 17p11.2 duplication in patients with axonal CMT (MNCV>30 m/s): approximately 2%, corresponding to a single family. In this family with dominant inheritance, only one patient underwent an electrophysiological examination, showing a median MNCV of 40 m/s but a latency of 6 ms and a reduced cubital MNCV of 33m/s. The duplication was also detected in the affected patient's mother, for whom no electrophysiological data were available.

Twenty-one unrelated patients with demyelinating CMT and without 17p11.2 duplication were analyzed by non-isotopic SSCP for the P0 and PMP22 gene. Five and two mutations were detected on the P0 (chromosome 1) and PMP22 (chromosome 17) gene, respectively, corresponding to 35% of the tested patients.

In order to evaluate the frequency of Connexin32 (Cx32) mutations in axonal CMT, we planned to perform a systematic screening of patients with axonal CMT belonging to pedigrees without male-to-male transmission, and of isolated axonal CMT patients. For 29 unrelated cases with axonal CMT, familial and isolated cases, presenting with intermediate median MNCVs (between 30 m/s and 40 m/s), the entire coding region (exon 2) of Cx32 gene was sequenced and 11 mutations were identified (38%). For the remaining families, only the half of the gene (261 to 722 bp) was analyzed. The frequency of abnormal SSCP profiles reached 30%, for both familial and isolated cases. According to the report of the European consortium on the distribution of mutations in the Cx32 gene, we can anticipate that the frequency of Cx32 mutations in axonal CMT will reach 40% to 50%.

The frequency of the 17p11.2 duplication according to the type of the CMT demonstrates the accuracy of the threshold value of 30 m/s for median MNCV to discriminate between demyelinating and axonal CMT. Median MNCVs were measured in patients from families with axonal CMT and without male-to-male transmission. Seven families showed missense mutations and three families nonsense mutations. Cx32 mutations were detected in 48 at-risk relatives (21 males and 27 females). Median MNCV is rarely lower than 30 m/s in CMTX patients.

■ OF INTEREST

St. John's wort, often known by its Latin name, *hypericum*, is one of the hottest herbs going. The herb is a remedy for depression. The respected *British Medical Journal* last year reviewed all the literature that has been published on hypericum, including 23 double blind studies, many in Germany. In Germany, St. John's wort is prescribed twice as often as Prozac-type drugs. Psychiatrist Allan Peterkin, assistant professor of psychiatry and family medicine at the University of Toronto says, "The Germans are pretty good scientists and there's good science behind this herb." Hypericum is not stimulating or addictive and is available without a prescription for about \$10 for a bottle of 60 300-milligram capsules. Hypericum works similarly to Prozac, inhibiting reabsorption of the brain chemical serotonin and producing feelings of well-being. The drug is not a panacea, doctors say, and should never be taken along with conventional antidepressant drugs. It is not recommended for severe depression.

(Editor's note: As with any new medication, consult your doctor before beginning this or any other treatment.)



OF INTEREST

Researchers have found a hormone that stimulates growth of the protective sheath around damaged nerves, a development they say could help in the treatment of some degenerative nerve diseases. Damage of the myelin sheath interrupts signals between the brain and nerves in nerve diseases like multiple sclerosis, diabetic neuropathy, and amyotrophic lateral sclerosis.

Experiments conducted at the University of Michigan focused on diabetic neuropathy, a severe condition in some diabetic patients that destroys the nerve endings in feet and can lead to amputation. Laboratory tests found that an insulin-like growth hormone produced by the liver called IGF-I lessened the damage from diabetic neuropathy on nerve cells in rats. Although several growth factors are being studied, IGF-I appears to be the most effective at inducing the growth of the sheath and preventing neural cell death.



Summer Research Fellowships:

Conclusion: a strategy for molecular diagnosis

According to the distributions of 17p11.2 duplication and Cx32 mutations in the different subtypes of CMT, we propose that the 17p11.2 mutation should be tested first in patients with median MNCV below 30 m/s, but when MNCVs are above 30, both Cx32 mutations and 17p11.2 duplications should be considered. Screening for Cx32 mutation should be carried out in isolated cases or in families whose members have intermediate or normal MNCV of median nerve with no male-to-male transmission.

Genetic Anticipation

SPENCER BLACKMAN, MD, PhD Student,
University of Miami School of Medicine

Major advances in the understanding of the molecular basis of a growing number of neurological disorders have been achieved through the identification of genetic defects in patients, as well as in animal models for these diseases. One of the neuromuscular diseases to which this statement applies is Charcot-Marie-Tooth (CMT) disease. CMT, also termed the hereditary and motor sensory neuropathies (HMSN), represent a clinically and genetically heterogeneous group of disorders. Great variability in clinical presentations exists even within families that possess the same genetic defect. The underlying mechanisms responsible for different disease manifestations are unclear.

We had previously identified four CMT families in which disease severity increases in succeeding generations (genetic anticipation). A Clinical Severity Scoring System (CSS) was developed to clinically evaluate these families. Using the CSS, a decade-wise global neuropathic deficit score was calculated for each affected member, as well as a longitudinal profile of the CSS over decades, reflective of the comparative severity and clinical course of CMT1A neuropathy among affected family members. Although statistical analysis of this information is still being completed, the CSS data appear to correlate with our clinical observations. In the past year and a half, we have identified four new families in which anticipation appears to exist. The CSS is being similarly applied to these families. This collection of these families offers a

unique experimental opportunity in which to address the key issue of variable phenotypic expression. Spencer was instrumental in the further ascertainment of these families, obtaining more detailed family and medical information, arranging collection of blood samples from the far reaches of the globe, and processing DNA samples for molecular studies.

The most common mutation in CMT1A families is the large duplication that consists of a 1.5-mB monomer unit flanked by large repeat segments termed CMT1A-REP in tandem array. This occurs in approximately 55% of CMT1A patients. The human pmp-22 gene, other genes, and several low copy repeat sequences, are entirely contained within this duplication. The remainder of the CMT1A patients display a variety of pmp-22 point mutations. Despite the observations that patients with the most severe demyelinating forms may have either a large 1.5-mB duplication in the 17p11.2 region or one of several reported point mutations in pmp-22, genotypes do not correlate with disease phenotypes in the collection of patients analyzed.

Our research continues to focus on the hypothesis that an increasing severity of disease presentations within kindreds is associated with DNA rearrangements that disrupt a number of normal genes in the CMT1A duplication region on chromosome 17p11.2, thus giving rise to different disease manifestations. This large DNA duplication has been correlated with the occurrence of symptoms typical of CMT I, the most common form of HMSN. Several previous observations suggest that alterations in the size of this duplication are feasible, and that repetitive DNA sequences within the 17p11.2 region are involved in subchromosomal DNA rearrangements and delineation of duplication size.

Our hypothesis continues to be tested through the detailed molecular analysis of the 17p11.2 region in genomic DNA of affected individuals from these families, assaying for the duplication mutation, as well as for an alternate mutation, a single-base-pair change in the gene for pmp-22 within this region. To date, the 1.5-mB duplication has been detected in every CMT1A family with anticipation analyzed. Our next immediate goal is to assay for alterations either in DNA size of the duplication mutation, or in DNA sequences surrounding the pmp-22 mutation using pulse field gel (PFGE) and restriction enzyme analyses.

A Review (cont.)

The aim of our continued studies is to complete the preliminary duplication studies and clinical evaluations in any remaining families, such that statistical analysis of clinical data can be completed in preparation for the next set of detailed molecular studies that are ready to begin. These studies are being generously supported through a 1997-98 postdoctoral fellowship through the CMT Association.

Another related important study in which Spencer was intimately involved this summer was the clinical and molecular analysis of African-American patients with CMT. We recently described (American Society of Human Genetics meeting, October, 1997) our first studies involving two unrelated severely affected HMSN patients of African-American ancestry. As a result of molecular investigations, a novel polymorphism in intron 3 of pmp-22 was detected in both patients. Analysis of control DNA samples revealed that this polymorphism was absent in Caucasians, and present in 35% of the African-Americans analyzed. It is of extreme interest that the intron 3 polymorphism was found in addition to a CMT1A locus mutation in two unrelated severely affected patients, suggesting a possible influence on disease phenotype. Spencer's efforts were essential to the identification, ascertainment, and recruitment of additional families into this study, which may prove to be extremely important to our understanding of the disease process in CMT.

We would like to end our report with these thoughts. Clinical variability has long been noted in the hereditary motor and sensory neuropathies, and at the present time, our understanding of disease genotypes does not correlate with disease phenotypes. This is one of the biggest unanswered questions in CMT research, as well as in family concerns. It is of extreme interest that the intron 3 polymorphism we have detected was present in conjunction with a CMT disease-causing mutation in two unrelated severely affected patients, possibly producing an additive effect (disease worsening). We are further investigating the frequency of this polymorphism, and possible affects on disease outcomes, in additional CMT patients of African-American ancestry.

This work could not have been completed without the continued support of the CMT Association, as well as the numerous physicians and families who have graciously consented to participate in these studies.

GIFTS WERE MADE TO THE CMTA

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Sarah Bergmann

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Ted & Mina Futterman's 50th Anniversary
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Rose Rosengarden's 90th Birthday
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Estelle Stokes
Marvin & Adele Greenwald

While a great deal of effort has been made to make these list as accurate as possible, we apologize for any omissions or misspellings that may have occurred.

Creating an Accessible Home

By **SHELLEY PETERMAN SCHWARZ**

Editor's note: This article is reprinted with the kind permission of the author. The article originally appeared in Enable, January/February 1998. Enable is the official magazine of The American Association of People with Disabilities. Shelley is severely disabled by multiple sclerosis and her independence in her home has been made possible by using inexpensive solutions to ordinary problems.

Shelley has recently expanded and updated her book, Dressing Tips and Clothing Resources for Making Life Easier. This unique, large-print reference shares hundreds of tips and lists more than 100 resources for easy-on/easy-off clothing for men, women, children, and wheelchair users. Mail \$19.95 plus \$3.00 for postage and handling to Shelley, 933 Chapel Hill Road, Madison, WI 53711-2405. Credit cards are accepted. Phone 608-274-4380.

Twenty-five years ago, my husband Dave and I decided to purchase our own home. We told the real estate agent we wanted a two-story house because we had both grown up in two-story houses. Then, on a dreary day in March, I saw an advertisement in the classified section of the newspaper about a ranch-style home in a neighborhood we liked. We took a look and decided to take a gamble and buy it. Looking back, it was a fortuitous decision, because I would later develop multiple sclerosis and a wheelchair-accessible house would be necessary. Over the years, we've had to make only a few major changes to our home to accommodate my increasing disability. We brought the laundry room up to the first floor. We ramped our sunken living room, and we put in a wheelchair-accessible shower.

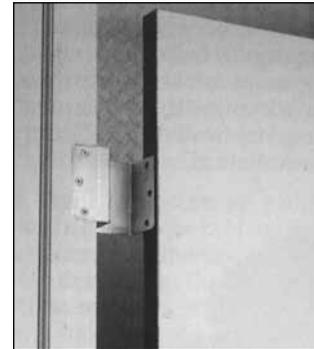
However, it's the simple modifications and devices we have made or purchased over the years that really give me my freedom and independence. Many of the modifications have little or no cost, and most of the devices can be purchased for well under \$100. The prices we have listed represent the minimum cost we found at either home improvement stores or in the catalogs listed at the end of this article. Don't be surprised if you find them to be a few dollars more.

DOORS, DOORKNOBS, AND DOORWAYS

Shortly after my diagnosis, I found it easier to grasp and turn a doorknob if someone wound several rubber bands around the largest part. When doorknobs finally needed replacing, we replaced them with lever-handle doorknobs (\$15 and up). Home improvement, hardware, and building supply stores have many styles to choose from. And always try out the handles before installing them to make sure they are easy for you to operate.

As an alternative to replacing doorknobs, you can put doorknob extensions (\$6) over your regular doorknobs. These devices create a lever handle, which makes the door easier to open. Installation is easy because the device fits right over a standard doorknob.

Doors that are hard to open because they rub on the floor or carpet can be adjusted without removing them. Put a large piece of sandpaper on the floor under the door and move the door back and forth a few times. You may need to put some newspapers under the sandpaper so



Offset door hinges are a cost-effective way to add one to two inches to a doorway.

there will be good contact with the bottom of the door. The sandpaper smooths the bottom of the door to prevent it from rubbing on the floor. As a result, the doors are easier to open.

For someone who will need a wheelchair long-term, a cost-effective way to widen doorways up to one or two inches is to install off-set door hinges (\$25). The hinges allow the door to swing out and away from the door opening. They are easy to install using the existing holes and screws, and they require no cutting or drilling. You can find the hinges at many hardware or building supply stores. If your wheelchair is too wide to get through your bathroom door, consider removing the door and doorjamb on either side of the door frame. Then, put up a tension rod at the top of the door opening and hang an opaque shower curtain liner (black) with curtain rings from the rod. This can be an inexpensive solution to a temporary problem. When you re-hang the door, oil the hinges.

Portable and roll-up threshold ramps will accommodate minor surface changes inside your house. Simply place the ramp over a standard or sliding door entrance. Ramps are made of various substances and will cost anywhere from \$70 up, depending on size and construction.

To solve the problem of unlocking and opening the heavy exterior front door on my house, I press the battery-operated garage door opener. The door to the garage opens, and I can use the garage entrance to my house. To prop open a heavy door, I use a tablespoon turned upside down. Push the handle under the open door.

LIGHTS, LIGHT SWITCHES, LIGHT BULBS, AND OUTLETS

Wall switch extenders (\$7) lower a light switch about 12 inches below the actual switch, making it easy to turn on and off from a wheelchair. Some extenders mount over a standard single light switch, while others replace the existing

With Projects Under \$100

wall plate using the same screws. The device is easy to attach and will not scratch or damage walls. Over time, we have replaced many of our traditional light switches with rocker-panel light switches (\$4) because they require less fine-motor control. Rocker-panel switches can be turned on by pressing with an arm, an elbow or the palm of the hand and come lighted or unlighted. You can find them at discount, building supply, or hardware stores. We've also installed a rotary dimmer switch (\$6) in our bedroom. When I'm resting, my husband can turn on a dim light without disturbing me.

Some other electrical devices that might prove helpful in illuminating your home are the following:

- "Touch" lamp controllers (\$7), which change the intensity of a lamp, generally in four steps, when a metal part on the lamp or control pad is touched (non-metal lamps require a device with a control pad).
- "Slide" (\$10) and "Rocker" (\$15) dimmer switches to control room lighting.
- "Infrared" motion detectors (\$12), which turn on lights automatically when you enter a room.

A vertical outlet extension device brings baseboard electrical outlets up to the wheelchair user or anyone else with reaching or bending limitations. No special wiring is needed; it just plugs into an existing outlet. A pull-plug device makes it easy for people with weak hands to insert and remove stubborn electrical cords.



A wall-switch extender brings light switches to a convenient level.

At my computer, I plug all of my computer equipment—the monitor, printer, and CPU—into a six-outlet power surge protector I bought at the hardware store. By pressing one switch, I turn on all my equipment at once. You can do this, too, when you have several items you want to turn on all at one time.



A tap turner makes using the sink easier.

FAUCETS AND SINKS

Washing my hands in the bathroom sink was difficult, because I couldn't stretch my arms out far enough to get them under the water spout. I didn't have this problem in the kitchen because the spout was longer. So, we replaced the short-spouted bathroom faucet with a long-spouted kitchen faucet.

A tap turner is a device that helps turn faucets on and off. Like the doorknob devices, the turners create a lever handle for easier grasping. Some tap turners can be attached with clamps; others just rest on top of the tap, while others are a portable hand-held device that gives the user additional leverage (UniTurner™ \$16). Two water controls, one for hot water and one for cold water, are easier to operate if wrist blades are installed. Wrist blades are wide, flat, wing-type handles that allow you to turn water on and off by pushing with the forearm, the wrist, or the heel of the hand.

Hospitals use institutional-looking faucets, but some plumbing manufacturers, such as Delta, offer decorative wrist blade fixtures. If necessary, install the faucet on the right side or left side of the sink. However, this change will be more costly because it will involve the replacement of the sink as well as the counter top. If installing new faucets, check out those intended for mobile homes and laundry areas, as many of their designs include longer spouts and/or wrist blades.

Shelley Peterman Schwarz can be reached by e-mail at help@MakingLifeEasier.com. Her web site is www.MakingLifeEasier.com

■ WHERE TO FIND THEM

Some of the products listed in this article are easier to find than others. If you can't find the products you need at a local home improvement, building supply, or hardware store, contact specialty mail order companies and ask them to send you a catalog. Most items mentioned here are carried by one or more of the following companies:

AdaptAbility catalogue, P.O. Box 515, Colchester, CT 06415-0515, 800-288-9941, fax 800-566-6678.

S & S Worldwide Opportunities catalogue, P.O. Box 513, Colchester, CT 06415-0513, 800-266-8856, fax 800-566-6678.

Sammons Preston's Enrichments, P.O. Box 5071, Bolingbrook, IL 60440-5071, 800-323-5547, fax 800-547-4333.

Independent Living Aids, Inc., 27 East Mall, Plainview, NY 11803, 800-537-2118, fax 516-752-3135.

OF INTEREST

Sammons Preston's *Enrichments* catalogue offers a wide range of products for special needs. The catalogue was highlighted at the Florida patient/family conference by the occupational and physical therapists and contains items such as folding canes, hand-strengthening devices, jar openers, exercise equipment, lightweight "reachers" and many other items that patients might find useful. The customer service number for Sammons Preston is 800-323-5547. Ask for a catalogue to be mailed to you.



Do You Need a Certified Pedorthist on

By **NANCY HULTQUIST, Director,**
Pedorthic Footwear Association

Footwear can be critical to foot health—yet relatively few patients experiencing foot problems realize they can get expert footwear help to stay as mobile as possible.

Pedorthics (ped-or-thiks) is the design, manufacture, modification, and fit of footwear, including foot orthoses, to alleviate foot problems caused by disease, overuse, or injury. Basically, pedorthics uses footwear, carefully fitted, to relieve or accommodate temporary or permanent foot problems.

Although it should seem both simple and logical to consider shoes as part of a patient's mobility equation, the reality is quite different. Many patients who wear a foot orthosis get only the most general type of instruction regarding the shoe they'll need to wear the orthosis effectively—e.g., "Get a good athletic shoe," or "You'll probably need a longer or wider size shoe with this." But, of the hundreds of athletic shoes on today's market, what constitutes a "good" one? More importantly, what constitutes an appropriate one for a particular patient? Should you look for a running shoe, a cross-trainer, a walking shoe? Does it make any difference? What if an athletic shoe is simply

out of the question for the occasion or event? What kind of "dress" shoe should you look for? Particularly among patients with some degree of reduced foot sensation, how can you judge appropriateness of fit?

Because most people are conditioned to think of shoes as "fashion statements," few of us think of shoes as a primary facilitator of function. Yet feet serve as a foundation for movement. Wearing a shoe that is inappropriate for your functional needs is like assuming a professional baseball player will play well with whatever glove he happens to put on... but would a wool mitten perform as effectively as a leather baseball glove?

If you have a footwear prescription, you probably need a certified pedorthist (CPed) on your health care team. Certified pedorthists are specialists in patients' footwear. They study shoe construction and modification, foot orthotics materials and fabrication, and lower extremity anatomy, physiology, and biomechanics; they understand that the foot, the orthosis, and the shoe must be viewed as a unit, or the patient's foot orthotics can't be expected to function as prescribed.

For individuals with CMT, footwear can be a critical element in their continuing ability to stay active. CMT has a wide spectrum of symp-

In a Sleepy Town...the "Sole" Provider

By **ARDITH FETTEROLF, CMTA Board of Directors**

Versailles, Missouri is a sleepy Ozark village, centered around a vintage courthouse square. Driving around the square, I noticed that there were many stores and most of the parking spaces were taken. Just off the square was my destination—Gollihars Shoe Store.

When I entered the store, it was apparent that it was more than just a shoe store. It is a meeting place where people are welcomed by Donna and Larry Clark, the owners. I stopped at Gollihars to see the Clarks and especially to meet Lydia Shirk and her daughter, Irene Sauder. Both mother and daughter have CMT, as does an aunt. The Shirk and Sauder families are of the Old Order of Mennonites and are among the 150 families who attend the Clear View Church. There are other Mennonite groups in the area; some drive automobiles and some uti-

lize horse and buggy transportation, as the Shirks and Sauders do.

Both the mother and daughter have CMT-related foot and ankle problems and wear special shoes adapted to their feet by the Clarks. Lydia and Irene were seen by Dr. Ian Alexander when they accompanied the Clarks to the Akron Ohio Crystal Clinic Conference hosted by the CMTA. At that time, the doctor thought they had all the classic symptoms, so no blood test was done. Irene wears a New Balance athletic shoe with a custom-molded stabilizing orthotic. Her foot problems have not increased much since she began wearing the orthotic. She requires a new pair of shoes each year, as she wears them constantly. Lydia wears a pair of PW Minor Depth-in-Lay shoes with custom-molded orthotics and specially designed stabilizers called flairs so that she can keep her balance.

Donna and Larry Clark, who is a certified

Your Team?

toms, especially as manifested in foot conditions. And, as anyone who's ever tried a shoe insert knows, the best insert in the world won't help unless you can wear it properly. That's what makes knowledge of shoes, their construction, and their possible modification so necessary.

Obviously, inserting any type of orthosis into a shoe takes up room that was originally designed to accommodate the foot. It should be equally obvious that an external shoe modification can contribute to a gait change, which in turn might affect another joint, requiring medical diagnosis and treatment. Shoes can also affect the dermatological condition of a foot, which may create a need for medical attention.

Most CPeds work primarily from a doctor's prescription, although they can provide over-the-counter footwear assistance to individuals who don't require a prescription. Because they can recognize routine foot conditions that may require medical attention, a CPed is often able to refer a patient to a medical caregiver before a condition becomes a problem.

Although there are only about 1,500 CPeds in the US today, the number of certified pedorthists is growing at a rate of roughly 20% a year. It's a still small group to address the needs of a large and diverse population, particularly

pedorthist, have owned the Gollihars Shoe store for 27 years, returning to the area when Donna's father passed away and her mother was unable to continue the store's operation. Donna checks patient's feet, massaging their legs and feet to help find the exact trouble spots. The couple decides where shoe and insert adjustments need to be made by Larry. Donna calls him the "mechanic" because he is a mechanical engineer in addition to being a certified pedorthist. Both of the Clarks gave up professional careers in education to return to Versailles to continue the tradition of solving shoe and foot problems in the family store. Larry has been a pedorthist since 1981, and each year the couple attends a national conference to update their knowledge.

Besides Lydia and Irene, the Clarks see several other families with CMT, some with several generations with the disorder. Their special shoe store serves a radius of at least 100 miles. They make adaptive shoes for people with many other walking problems.

as the baby boom generation steps into middle age and encounters the problems associated with aging, arthritis, sore feet, and/or the consequences of five decades of choosing footwear primarily because of its appearance instead of its function.

But if footwear—both shoes and orthoses—can be part of the solution to your foot problems, please ask your primary health-care provider to add a CPed to your health care team. Choices about staying as active as possible shouldn't be left to chance.

For a list of CPeds in your state, please send a self-addressed, stamped (32 cents) envelope with your request to: Board for Certification in Pedorthics (BCP), 9861 Broken Land Parkway, Suite 255, Columbia, MD 20146-1151. BCP is the institution that sets the national standard for the practice of pedorthics and tests individuals to determine whether they meet entry-level standards.

Information for this article was provided by the Pedorthic Footwear Association, a non-profit membership association of individuals engaged in the practice of pedorthics; in manufacturing, development, and distribution of pedorthic devices; and in research and education on the topic of pedorthics. PFA is also located at the Columbia address listed above.

Lydia and Irene, in their black bonnets and knit dresses, touched a special place in my heart and the Clarks made my first visit to a pedorthist very rewarding and informative.

Larry Clark, certified pedorthist, works on a custom-made insert in his Versailles store.



A Journey Toward Wellness, Part II

By CYNTHIA GRACEY

A place to begin: become aware of your personality type

Part I of this article appeared in the Fall 1997 issue of The CMTA Report.

While I did not begin my own journey with a working knowledge of the distinctions of personality type, I wish that I had. I would have trusted my decisions, taken more “correct” turns, and been able to share with those who so wanted the information I was collecting in ways that they could more fully comprehend.

When I began to share with others my rather different journey towards enhanced well-being, vitality, and (yes) better mobility in my arms and legs, I naively thought that if someone just did what I did and followed in my footsteps he or she would have similar experiences to my own.

I dismissed many people who were sincerely trying to find their own alternative route to greater vitality but who resisted or misunderstood “my way” as just not really ready to get well or needing their symptoms for some unconscious reason that I was not qualified to fathom. What I have learned in the most positive sense is that for the most part those dear seekers of a healthier life were probably just not my “TYPE”.

Several years ago I was introduced to the profound benefits of an awareness of my own personality type. This body of knowledge about personality type was first articulated by Carl Jung and then made accessible through the pioneering work of the mother and daughter team of Isabel Myers and Catherine Briggs. Over a period of 40 years, with the research and revisions still continuing to this day, they devised what is widely known as the Myers Briggs Type Indicator. At its worst, this work has been utilized to pigeonhole persons in inappropriate ways. At its best, it offers insight into the diversity of how we each process information, decide, learn, energize, revitalize, restore, organize, become motivated, listen, and communicate.

Had I known of the existence of this work and made use of it, I would have saved much time, heartache, frustration, self-recrimination, guilt, anger, sadness, etc.—all detrimental for one navigating toward enhanced well-being. I also would have been more of a positive influence for those who crossed my path seeking guidance.

As a way of general reference, the Myers Briggs Personality Type Indicator offers clarity, perspective, understanding and research on the following:

- How individuals best 1) get energized, revitalized, handle stress, restore to equilibrium, and 2) best process their ideas and information
- How individuals do their best perceiving, “taking in” and processing of information, and what kind of information is most likely to be considered important, relevant or simply noticed.
- How individuals make decisions and effectively use the information they have gathered.
- Whether a spontaneous or an ordered system is preferred in an individual’s outer life and likewise for his or her inner life.

Awareness of the above, forms the basis for categorizing individuals into 16 possible combinations of preferences called “types.” The categories that you self-select demonstrate your preference for one of the following:

Introversion, energizing alone or with a significant other (friend, child) and processing ideas internally over time, or,

Extraversion, energizing with others and processing ideas best in conversation with others.

Sensing, gradually collecting detailed information through your five senses.

Intuition, immediately taking information and forming it into patterns of information.

Thinking, decision-making based on a logical and analytical system of rationality.

Feeling, decision-making based on a complex system of rationality based on a gestalt of values, nuances, personal emotions, timing etc.

Judging desire for completion and decisive action.

Perception preference for open-ended possibilities and process.

The above descriptions are simplified explanations, which would be analyzed by a qualified MBTI consultant. Awareness of “type” has been helpful to me, both personally and professionally. For instance, I know that...

The environments that support my well-being, those that nurture and revitalize, provide sanctuary for healing, and restore balance, correlate with my type. I prefer extreme introversion and therefore require much more solitude and total alone time than most other people. I have also become aware that my type of “healing

sanctuary” could prove, over an extended time, to have an adverse effect for certain other “types” particularly those who are more extroverted.

I am drawn to certain processes: solitary meditation, visualization, extensive reading alone at night—all of which are effective for me but may not be for others.

Certain stress inducers affect my vitality and wellness and are very type related. My least preferred functions are Sensing and Thinking and when I have to do too much of either I break down and my physical well-being suffers. My body is the barometer of my ability to balance the stress in my life, so this knowledge is critical for me.

Specific suggestions and solutions can be designed for interaction with care givers, family members, and friends. For example, I like silence while being massaged or having any body work done and like to have any impressions given at the end of the session. Expressing this improves the interaction.

The data compiled regarding personality type make it possible to project the areas of potential challenge and provide possible solutions as individuals find their own unique course towards greater vitality.

Personality type and maintaining a total wellness program of fitness, nutrition, and exercise are related. Someone whose preference is routine as opposed to spontaneous would require a different approach to wellness.

With regard to life’s choices, I generally gather information and then make decisions through a particular interplay of processes, identifiable through the MBTI. Effective mechanisms for decision making lead to better decisions, which in turn lead to improved well-being. Remember that the MBTI personality assessment is the best recognized, best researched, and has the most informative body of knowledge. There are other personality type assessments out there.

I have personally found useful Bernice McCarthy’s work on learning and leadership styles and the Enneagram.

The MBTI is now used in large corporations, in career suitability and relationship counseling, in team trainings, in personal development, education, and religious institutions. I am probably one of the few if not only professionals to use it in connection with the suitability of well-being choices.

You may have done the MBTI assessment and discounted the profound impact the information could have on your physical well being. So, if you are serious about walking down this “not quite mainstream” road to well-being, I hope you find a qualified person in your area to administer the MBTI or contact me.

How each of us chooses to walk our path, that includes physical challenges, must be individually tailored. There is no right or wrong way, only the way that works for each of us. Understanding our unique differences, our different needs, our different values, our different ways of learning, listening, communicating, deciding and then making all that work to effect positive changes in our physical well-being is no small feat, but I have found it well worth the effort. I strongly encourage you to explore this avenue of self-awareness and personal discovery.

I would be delighted to answer specific questions. If you e-mail or write to me, please let me know if I can share your e-mail or street address with others of similar interests and if I can use your questions or comments in any upcoming articles. Contact me at PATHCG@aol.com or 1109 Harbor Drive, Delray Beach, FL 33483.

Editor’s note: Space restrictions prohibit inclusion of the entire article, but Cynthia’s work can be found at the CMTA website: www.charcot-marie-tooth.org

New Medical Equipment Exchange Program

The National Organization for Rare Disorders (NORD) has announced a new program called Medical Equipment Exchange. The program’s purpose is to provide people with inadequate health insurance with the means to purchase needed medical equipment. Items such as hospital beds, wheelchairs, toiletting equipment, and even canes and crutches would be included. Information about this new program is available through NORD’s home page at <<http://www.NORD-RDB.com/~orphan>> or by writing to NORD, P.O. Box 8923, New Fairfield, CT 06812. NORD will act as the program’s facilitator and assumes no responsibility for the quality, performance, or medical results of the equipment. People with used medical equipment are urged to send their ads to NORD via the Internet, e-mail or regular mail.



CMT on the Net: International CMT Chat

This is the first of a series of articles pertaining to CMT information and support resources on the Internet. Readers are encouraged to contact Stephen Scofield (CMTA Board of Directors) via e-mail at SScofield@inerconnect.net with suggestions and comments. Look in future CMTA Reports for articles on the ABCs of getting on the Internet and the CMTA's coming ListServ distribution service.

Some of the "friends" who went "above and beyond" are, left to right: Dr. Reid Sutton (Genetics Fellow), Sylvia Brito Silva, Cristiane Brito Silva, Kim Penrod, and Dr. James Lupski.

Pretty much every evening, and often during the day, you will find a group of friendly people gathered in the Charcot-Marie-Tooth Chatroom on the Internet. The "chatroom" is one of approximately 50 created and managed by John Lester of Massachusetts General Hospital and provided as a free public service by MGH's Neurology Department. The Internet address is: [neuro-www3.mgh.harvard.edu/interaction\\$/chat/char](mailto:neuro-www3.mgh.harvard.edu/interaction$/chat/char).

Through meetings in the room, many close friendships have been formed among people, most of whom have never met in person, but who share a knowledge of CMT that is beyond medical wisdom. Discussions might center on how each of us is living with CMT, medical news, a new scooter, AFOs, shoes, canes, surgery, the zoo, kids, pizza, or WalMart. Sometimes, discussions are serious. Sometimes we "party." On Halloween, everyone came in costume. Around the holidays, many seasonal cards (electronic cards) were exchanged via the Internet.

Louise Smith, past member of the CMTA Board of Directors, whose talents and drive contributed much to the existence of the room, commented, "You can be completely anonymous if you wish and ask questions or make statements you wouldn't anywhere else. There is access to people any time of day or night, and all over the world. I particularly enjoy speaking to the people on the other side of the world. CMT is rare; the chatroom removes the isolation."

Recently one of the CMT support groups in Australia had an idea pertaining to the chatroom

that is described below by Diane and Bill Lean from Adelaide, Australia:

The idea for this unique experiment evolved during one of the regular CMT support group meetings at the MDA in Adelaide. It came as a result of the need for something different and interesting to be made available, as the group was having difficulty finding content for their meeting. As we are Internet "freaks," we thought it would be a good idea to arrange a "virtual" meeting with the "regular" group from the MGH chatroom. Once we contacted the group and overcame the "time zone factor," we were able to set the whole thing in motion. On the day we had loads of fun with up to 11 in the chatroom and in Adelaide there were nine of us keeping Bill typing frantically. We know everybody found it useful comparing "notes" on a wide range of topics relevant to their CMT. It is planned to repeat the exercise on a regular basis so that some friendships can be made by people with a common interest.

Your first time in the chatroom you might run in to some of the regulars: Amy, Barb, Believer, Beth, Bevy, BillinOz, Bren, Cactus, Callie, Candie, Capo, Carol, Char, Connie, Dan, Darlene, Didi, Elena, Everready, Goofy, Goon, Gumby, Heidi, Holly, Janet, JaRae, Jewelee, Joni, Kat, Kim, Louise, Lulu, Mary, Numby, Paige, Pat, Paul, Peek, Rick, RonD, RonS, Ruthie, Scootz, Sekhmet, Shepherd, Sherry, Smoky-Joe, Stephen, Stuart, and Vaughn. Collectively, we represent Canada, the United States, Australia, New Zealand, England, and Sweden.

You can be sure of a warm welcome.

—Stephen Scofield



ABOVE AND BEYOND

Once in awhile, large groups of people work in concert to make wonderful things happen. This issue's Above and Beyond effort was just such an instance.

In May of 1997, a Brazilian physician contacted Dr. James Lupski at Baylor College of Medicine in Houston to see if his clinic would evaluate a 15-year-old young woman, Cristiane Brito Silva, from Rio de Janeiro who was tentatively diagnosed with Charcot-Marie-Tooth disorder.

Because the family has limited financial resources, American Airlines (the teenager's mother works for the company) donated the air-

James A. Freaney (1916-1998)

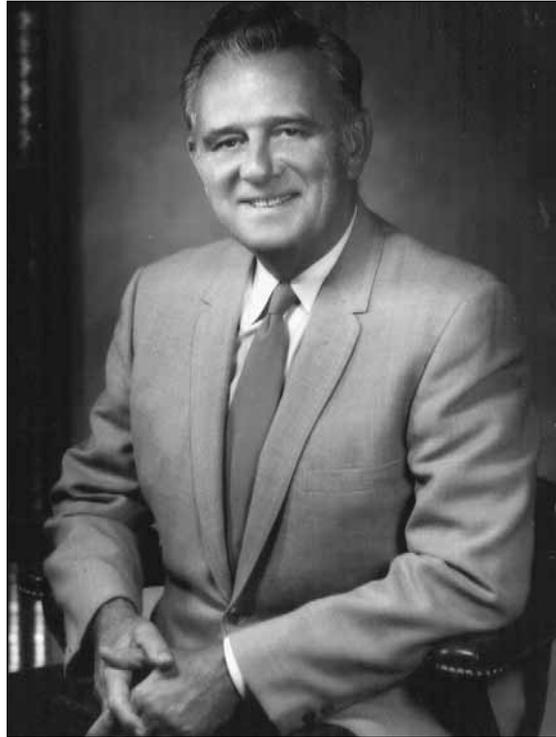
Loving Husband, Father, and Friend of the CMTA

James A. Freaney was born in Boston, Massachusetts on August 6, 1916, the only son of an Irish Catholic couple. Growing up in the Depression, he took his responsibilities to his family very seriously. He helped support his family through odd jobs and a variety of entrepreneurial ventures. He graduated from Milton High School in 1934 and gave up college because there was no money. He worked as a bell hop, fell in love with Dot Biddle on the beach in Florida, and continued their romance through to their marriage in 1941. He made a living by selling fur coats door to door during the Depression until he took over his father's trash collection business in Boston.

Although he had only a high school education, Jim became well known as an expert in solid waste management, becoming an Associate Professor at MIT. He helped author the book, *The Treatment and Management of Urban Solid Waste*.

In 1977, he and Dot retired to Florida, where he enjoyed walking on the beach, dining and dancing, studying the stock market, and learning the computer. He took care of his beloved wife, Dot, for 56 years. His infectious sense of humor and wonderful Irish gift for story-telling will be remembered by everyone who knew him.

To keep his memory alive and to further research on Charcot-Marie-Tooth disorders, which affects his wife, his daughters, and his



James A. Freaney
August 6, 1916- January 8, 1998

granddaughter, the family has established the James A. Freaney Memorial Fund. The fund will support a summer research grant given annually to a student studying some applied or genetic research topic specifically related to CMT.

GIFTS TO THE FUND IN MEMORY OF JAMES A. FREANEY:

His wife,
Dorothy M. Freaney
His daughters,
Diane M. Freaney
Donna M. Freaney
Linda A. Freaney
His granddaughter,
Allison E. Harter
His grandsons,
Ben Suscavage
David Suscavage
Sam G. Lee
Wanda C. Hagan
Mrs. Minnie K. Slayter
Ardith Fetterolf
Mr. & Mrs. Burt Reed
Sue B. Cushman
Margaret G. L'Hommedieu
Carol & Curtis Clapham
Robert & Judith Donovan
Ruth & Herbert Ruhr
Paul R. Flynn
J. Rodman Steele, Jr.
Juliet J. Goodfriend
Ed and Betty Grieves
Nancy and Edward Hopey
Eleanore Gately Farrell

fare for them to travel to Houston. A representative of American contacted the Baylor clinic by phone and by fax to make arrangements for all the needed appointments.

Dr. James Killian, a neurologist with Baylor College of Medicine, who works closely with Dr. Lupski in evaluating CMT patients, kindly agreed to perform nerve conduction velocity and electromyography studies at no charge for the girl's family.

Because of findings reported by Dr. Killian, Dr. Lupski requested DNA studies on Cristiane for spinocerebellar ataxia (types 1,2,3,6, and 7), all of which were performed by the Baylor DNA Diagnostic Lab at no charge. The Baylor Biochemical Genetics Lab performed other studies, also at no charge. Baylor's genetics clinic paid for a vitamin B12 level.

Of course, Dr. Lupski donated all of his services for Cristiane.

A local resident, Kim Penrod, who happens to be a "friend of a friend" of the family, provided transportation and hospitality in her own home for Cristiane and her mother. Kim also happens to speak Portugese, which is the native language of this family, and she accompanied the family to the various appointments, providing translation service and lots of "comfort."

Pat Dreibelbis contacted Vince Bertolino at Athena Diagnostics who generously agreed to fund the connexin32 gene mutation study for Cristiane.

Through the unselfish generosity of so many people, Cristiane and her mother, Sylvia, made the trip to Houston and found out that she has CMT Type II. They returned home to Brazil a few friends "richer."

■ OF INTEREST

Thursday nights at 9 p.m. EST chat at The Disability Resource on "Parent's Rights and Responsibilities" and "Changes in IDEA and how they affect you."

Please join us Thursday night, 9 p.m. EST at <http://www.disabilityresource.com>
Just click on "Chat" link and sign in!



When A Child is Diagnosed with CMT...

Many parents report that the diagnosis of Charcot-Marie-Tooth disorder was just the beginning of a whole range of emotions they would eventually experience. Their emotions don't follow any set pattern or even last a predictable amount of time. They are, however, honest reactions and valid responses to the family's own personal circumstances. The father may be very accepting while the mother is very angry.



The important thing to remember is that all parents experience some emotions regarding their child's diagnosis and development. It is healthy, normal, and very common to feel angry, afraid, guilty, and sad. Once a parent can identify and acknowledge his/her feelings, he/she can begin to use some of the coping strategies that follow.

DENIAL OR DISBELIEF

Some parents say that they had a feeling, long before the diagnosis was finally made, that something was wrong. In many cases, with an

hereditary disorder, it is almost a situation of waiting for what they most fear: the same diagnosis that plagued their own youth. But, until the confirmation of their fears, they denied those feelings. This type of reaction is a very healthy emotion. It gives the person time to accept, to adjust, and to make plans. It can also protect a parent from too much pain, too soon.

SADNESS AND DEPRESSION

Many parents say that they experienced depression when their child was diagnosed with CMT. In some cases, if the child is the first family member to be diagnosed, the depression is a direct result of the ignorance of how serious the condition might become. Sadness can be tied into the dreams and expectations that the parents had for their child's life and their own. Often, depression or depressed feelings may occur unexpectedly, without the person's even knowing why. Seeing other children doing something that your child cannot do or cannot do without pain and difficulty can trigger long-hidden feelings of sadness.

ANGER AND HOSTILITY

It is not uncommon for the parents to direct their anger toward the doctor who diagnosed

Strategies for Coping

Cognitive Coping

- Read everything you can on your child's disability
- Talk to the parent of a child with a similar diagnosis
- Subscribe to newsletters on CMT or magazines like *Exceptional Parent*
- Attend workshops and conferences on CMT

Physical Coping

- Cry, laugh, have a sense of humor
- Keep a journal in which you write down your emotions and the coping mechanism that worked
- Exercise
- Eat well
- Rest sufficiently
- Take long walks
- Keep a normal routine

Psychological Coping

- Join a support group or start one
- Take up your child's cause, by raising money or raising awareness
- Count your blessings
- Take one day at a time
- Replace negative thoughts with positive ones
- Mentally list all the wonderful things your child can do
- Realize you're not alone
- Get counseling, if needed
- Find a parent who has experienced a similar situation

Spiritual Coping

- Talk to your family priest, rabbi, or minister
- Read devotionals
- Read the Bible
- Spend time in a peaceful setting: the woods, beach, or by a lake

their child, or toward each other, toward the child, God, or close friends and family. Often, they will hear themselves saying, "It's not fair" or "Life isn't fair." It is perfectly normal to feel anger at the unjustness of having a disability.

FEAR

Fear is a common reaction to the unknown. When parents are told that their child will not be "typical", they feel apprehensive. When that general fear is coupled with the lack of a clear picture of what the future holds for someone with CMT, because of its variable nature, parents are often doubly fearful. It is normal to feel anxious about the uncertain future, to feel inadequate as a parent, to shield your child from pain and unhappiness, and to feel poorly prepared to meet your child's needs, whatever they might be.

LEARNING TO COPE

Undoubtedly, if you have a child with CMT, you have felt some or all of these emotions.

Acknowledging your feelings and recognizing that other parents feel the same way will help you move toward accepting your child's diagnosis and planning your child's future.

It is when a parent gets "stuck" in a particular emotional state, unable to move on, that feelings become counterproductive. Most parents have developed coping strategies to help them "move through" their different emotions and get on with other things. On page 20 are some coping strategies parents have used to help them deal with the emotions they feel in response to their child's diagnosis.

Emotions are healthy responses to difficult situations. Using specific coping mechanisms will help you maintain a healthy balance in your life and allow you to deal with each situation in a productive and positive way. Emotions do not magically "disappear" or go away at some point in time. Feelings occur throughout your life as you encounter new and stressful situations, make difficult transitions, or realize that your child has missed out on some milestone you had envisioned for them. At those times, it is important to acknowledge your feelings, allow yourself the permission to feel that way, and reach into a "bag" of coping strategies to help you deal with the situation. Then, you can, again, get on with your life.

Editor's note: If, as a parent of a child with CMT, you have found any innovative coping mechanism to be quite successful, please write and let us know. We will share your story with our readers.

CMT Support Groups

Alabama/Greater Tennessee Valley

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

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, 707-253-0351

Florida/Boca Raton to Melbourne

Place: Columbia Medical Center, Port St. Lucie
Meeting: Quarterly
Contact: Walter Sawyer, 561-336-7855

Massachusetts/Boston Area

Place: The 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

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 Center
Meeting: Quarterly
Contact: Carole Haislip, 314-644-1664

New York (Westchester County)/Connecticut (Fairfield)

Place: Blythedale Hospital
Meeting: Monthly, Saturday
Contact: Kay Flynn, 914-793-4710

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

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

West Virginia/North Central

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

New Support Groups Forming!

Robert Budde will hold the first meeting of the Kentucky/Southern Indiana/Southern Ohio area CMT support group on April 4, 1998 at 1:30 PM at the First United Methodist Church in Lexington, KY. The first meeting will be organizational in nature and all interested persons should contact Robert at 606-255-7471.

On April 23, 1998, Dot Cain will host a meeting of a new Ohio support group at the Church of the Brethren in Greenville, Ohio. The meeting will begin at 6:00 PM and the group will meet on the fourth Thursday of the months April through October. For more information, contact Dot at 937-548-3963.

A third group will be forming in Flint, Michigan, under the leadership of Debbie Newberger and Brenda Kehoe, both registered nurses. The meeting will be on March 25, from 3-5 PM at the University of Michigan. For more information, call Connie Creech, RN, (health services) at 1-810-762-3456.

Members living in the Baltimore/Washington, DC area interested in a support group are urged to contact Michael Tapp at 410-760-1428. He is currently working to get a new group started there.



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

Ask the Doctor

Dear Doctor:

I am having multiple stress fractures in my metatarsals and one in my left tibia. Two orthopaedic surgeons, a rheumatologist, and a neurologist have concluded that these are due to CMT; however, I haven't been able to find any material that indicates a correlation between stress fractures and CMT. Is this common to the disease? —S.V.

A podiatrist from the Medical Advisory Board replies:

There is no direct association between CMT and stress fractures. CMT is an inherited neurological disorder and does not cause direct weakness or "fragile" bones. The disease causes muscle weakness and imbalance due to the nerve destruction. People who are active and walk or stand a lot put tremendous stress and strains on their bodies. With muscle weakness and imbalance, the bones of the feet and legs can become fatigued and thus develop "stress" fractures. These fractures occur in people who are not affected with CMT. The increase in activity and stress load on the bone causes the fracture, not the CMT.

Dear Doctor:

My wife has CMT. She has orthostatic hypertension, headaches, dizziness, and nausea most of the time. The doctors blame these problems on CMT. Are any of these symptoms normally caused by CMT? We do not know where to turn and the doctors we have seen so far cannot help. —W.M.

A neurologist from the Medical Advisory Board replies:

A full reply cannot be provided without review of the medical file of the woman. The symptoms mentioned are not usually regarded as complaints primarily related to CMT, and other more pertinent brain and nerve sources should be considered. Orthostatic hypertension may be due to an autonomic (nerves providing tone to the blood vessels) neuropathy, which is uncommon in CMT. A common cause is diabetic neuropathy, and if the woman is taking medication for hypertension, this can sometimes have an excessive effect and may need adjustment.

Dear Doctor:

I am interested in learning more about the CMT neurotoxic drug list. Why is a drug placed on the list; what is a megadose; and should we avoid all multivitamins with B6?

The Doctor replies:

The neurotoxic drug list was compiled by neurologists and is continually monitored by them for additions and corrections. A megadose is defined as ten or more times the RDA (recommended daily allowance). A daily multivitamin capsule should not be a problem. (See question and answer below.)

Dear Doctor:

I'm interested in finding out about the harmful side effects of pyridoxine (B6). I would like to know what B6's negative side effects might be. I've heard that a natural form of B6 may have a different effect than a synthetic form.

The Doctor replies:

Patients with CMT should have concerns about the harmful side effects of pyridoxine, vitamin B6. Although the lack of vitamin B6 has been associated with polyneuropathy, both experimental studies in animals as well as studies in humans have reported that excessive doses of pyridoxine produce a sensory neuropathy. Doses of 500 mg or more per day can result in a predominantly sensory neuropathy due to the degeneration of sensory neurons. It is not likely that pyridoxine itself would cause weakness or motor involvement. Purified pyridoxine has been used in the studies. Any of the natural foods that contain pyridoxine would have the same chemical, but they have other ingredients as well. I would suggest that patients with CMT use only small doses of pyridoxine such as the recommended amounts of 2.5 or 5 mg a day.

Classic Symptoms of CMT



"Classic presentation" of high arches and claw toes.

View of hand showing muscle wasting and contractures of the fingers.

Letters to the Editor:

Dear CMTA:

Besides the loss of mobility and use of my hands (which has deteriorated rapidly in the past two years), I have also experienced a lack of concentration, memory loss, and difficulty in getting my verbal message across to the listener.

As I read the articles in *The CMTA Report*, most of the focus is on the physical aspect of CMT. I wonder if there are others who, like me, are having this difficulty? If so, how are you dealing with this?

If anyone in the South Jersey area is interested in a support group, perhaps you could contact me and a group could be formed. Please fax me, Bill, at: 609-886-7312.

—B.W., New Jersey

Dear CMTA:

I am a 26-year-old female who was diagnosed with CMT at 21 years of age. A podiatrist I saw recently “strongly” suggested that I have surgery to perform a tendon transfer in both feet (move the tendon from the interior side of my ankles to the exterior side). He insists that he doesn’t want to fix anything, just make me more stable. I don’t fall or trip very much; however, my balance isn’t very good. He says he can still get me into a neutral position by taking out a wedge of heel bone and fusing my big toe. All this should make my gait more normal and a little stronger. I currently have pain from time to time, although it’s not unbearable and I wouldn’t want to make anything worse, obviously.

I plan on getting a second medical opinion; however, I would like to hear from others that have gone through this surgery, good or bad, so that I can make a more informed decision. Please write S.L. at the CMTA office or e-mail me at leasure@ix.netcom.com with your experiences or comments.

—S.L., California

Dear CMTA:

Hi, my name is Jon Ferner. I am 13. I go to Naples Central School. I am the only one in my school and town who has CMT. I was wondering if there was somebody else that would like to be pen pals with me. Maybe if we could all send in our ages and addresses, we could write to each other. I feel we could all use a support group. I

have many suggestions and could use some myself. My interests are ALL SPORTS. I am able to play sports now and I hope I will always be able to.

Editor’s Note: If any “kids” are interested in being part of a pen pal network, please send your name, address, and age to the CMTA office and we will prepare a pen pal list for distribution to everyone who responds.

Dear CMTA:

I have just received your literature. I thought you might be interested in my case. I am a retired high school French teacher. It seems ironic that two of the three scientists who gave their names to this affliction are French. I taught in Pennsylvania for 18 years, then married and moved south.

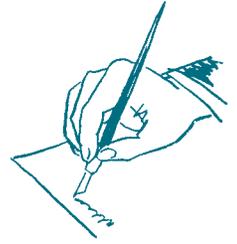
Several years ago, a local doctor diagnosed me with CMT. I was very frail as a young man, but had no physical disability until about 30 years ago. I began to have a leg weakness, mainly in my thighs. I have progressed very slowly, and today can still bathe and dress myself, although with aids. I can walk, but if I do not have “touch” holds, I use a walker that is specially designed for me with small wheels in front and nylon skids in back. I have a ramp to the rear door with two-inch steps and handrails. I have a chairlift to the upstairs. I do 15 minutes on a treadmill daily. I have no other physical problems and can eat and drink anything.

I cannot rise from a seated position without a handhold on something. I use the countertops in the bath and kitchen, a post in the bedroom, and the walker in the living room. I do not use a cane. I use reachers because I cannot bend over to pick things up. I cannot get up after falling. My wife has been my mainstay. I still drive, but not without my wife along. I drive only around town and only with a cellular phone.

For your information, I was 82 last November.

—F.C.M., Georgia

Editor’s note: This letter was reviewed by a member of the Medical Advisory Board. Considering the information given, with the predominant proximal (thigh) muscle weakness, there could be strong reservations about the diagnosis of CMT. The letter writer is recommended to consult a doctor in his area who is experienced in Charcot-Marie-Tooth disorders.



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.

The CMTA Report

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

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



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