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The CMTA Report

FEBRUARY 2001

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

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Working Toward a Cure: CMTA Awards Record Number of Post-Doctoral Grants

For the first time in the history of the organization, the generosity of the CMTA's membership has enabled the association to award five full-year post-doctoral grants to a diverse group of researchers from around the world.

JAMES THOMAS MOORE GRANT AWARDED TO AUSTRALIAN RESEARCH TEAM

The family of James Thomas Moore has chosen to support a clinical study which will investigate the use of night splints to stretch the long and short flexors in the leg and foot. The grant will be known as the James Thomas Moore Grant of the Charcot-Marie-Tooth Association. The recipient is Anthony Charles Redmond, working at the University of Sydney, Australia, under Professor Robert Ouvrier and Dr. Garth Nicholson of our Medical Advisory Board. The title of the project is "A Functional Approach to Calf Flexor Stretching in CMT1A: A Randomized Controlled Trial."

The understanding of the etiology and pathology of a wide range of neuromuscular diseases has improved substantially in recent years. In terms of the practical management of the progression of the diseases, however, approaches to the rehabilitation aspects of the disease process have not been so rapid.

The weakness and acquired deformity associated with CMT are significant causes of suffering, but no quality, randomized trials have evaluated any therapies aimed at addressing

either the foot deformity or the associated symptoms. Given that changes occurring in the lower limb are acquired over time, it is possible that a coordinated approach to understanding underlying muscular imbalances may improve the prospects for devising interventions to reduce the severity or rate of changes. Early in the disease process, exploration of treatments aimed at slowing the rate of progression and reducing the severity of end-stage deformity should be a priority for researchers. This study proposes to evaluate the effectiveness of addressing the underlying imbalance through a program of ankle splinting aimed specifically at opposing the pattern of changes seen in the disease. For the first time, this study will use a set of rigorous and quantifiable mea-

ures of the structure, function and health status of the patient with CMT1A to evaluate in detail the impact of the imbalance and any changes resulting from the treatment.

The program team in Sydney comprises two internationally known CMT specialists, a rehabilitation staff, a surgical staff, and specialists in a range of other disciplines providing expertise in genetics, internal imaging, biomechanics, anatomy and biostatistics.

The effect of a three-month period of night splinting on the structure, function and health status of the foot and leg in CMT will be evaluated by analyzing changes in a range of quantitative measures. The treatment approach will address the functional aspects of CMT foot deformity (i.e. the muscle imbalance and soft tissue contracture). It is believed that a combina-

(continued on page 2)

Reducing the severity of end-stage deformity should be a priority for researchers.

The CMTA.... Living our Mission

Mission Statement:
To improve the
quality of life for
people with CMT.

Vision Statement:
Dedicated to
generating hope.

GRANTS

(Continued from page 1)

tion of flexor-muscle overpull in the presence of extensor weakness explains much of the process of deformity. In this study, combined stretching of the long and short flexors in the leg and foot will be achieved through the application of dorsiflexion night splints for a period of three months.

The Moore family is pleased to support this practical study aimed at studying foot and leg weakness and deformity as a means of helping people currently affected by CMT.

THE REORGANIZATION OF THE AXON MEMBRANE IN ANIMAL MODELS OF CMT

For the second year, Dr. Edgardo Arroyo, working with Dr. Steven Scherer at the University of Pennsylvania, is a recipient of a post-doctoral award to study "The Reorganization of the Axon Membrane in Animal Models of CMT."

He will continue to study the organization of the axonal membrane protein in normal nerves and to compare that to the re-organization of the axonal membrane in mutant mice. Although the type 1 forms of CMT are demyelinating in nature, it is the axonal loss and not the demyelination *per se* which causes the important clinical deficits. Thus, understanding how demyelination leads to axonal loss is key to understanding the pathogenesis of CMT. It has been demonstrated that when the number of motor axons innervating weak muscles is severely reduced, age-related weakness and atrophy result.

Dr. Arroyo hopes to determine whether the extent of axonal membrane reorganization is related to the degree of demyelination and remyelination and to determine the time course during which the axonal membrane reorganizes. This understanding of how demyelination results in axonal loss will help with the therapies that are developed to treat the resulting weakness.

BOSTON UNIVERSITY STUDY: FETAL GENE THERAPY

Dr. Willmar Cadavid, working with Roger Lebo, PhD, FACMG, will conduct a study on the use of gene therapy in pregnant female mice. These investigators believe that prenatal gene therapy promises to be a safe and effective approach to prevent the manifestations of many diseases, including CMT. Prenatal gene therapy for CMT would correct disease symptoms by introducing normal genes or modifying only abnormal genes in the patient's own cells. Gene therapy is first tried in animal models. The protocols which produce the most favorable results are then tested in larger mammals prior to human testing.

Prenatal gene therapy has the potential to deliver normal genes to genetically abnormal cells to correct the abnormal genetic phenotype without disturbing other normal gene functions in the targeted cells. With the rapid advancements in prenatal genetic testing through amniotic fluid testing and chorionic villous sampling, fetal gene therapy is the logical extension of fetal diagnosis.

Fetal gene therapy would be able to deliver treatment early enough to prevent disease manifestation and tissue damage, especially for diseases with early onset. It is possible that the disease could be permanently corrected with one treatment if the gene vector could be integrated into the stem cell populations of organs or cell systems. For these reasons, this study holds great promise for future generations in CMT families.

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86 Combined Years at The Neurological Institute



Dr. Robert Lovelace, Chairman of the CMTA's Medical Advisory Board, attended a presentation on a new device for improving walking for CMT patients. With Dr. Lovelace is Dr. Daniel Sciarra, who is the senior neurologist at The Neurological Institute, New York City. He has been in practice and teaching at the Institute for over 50 years. Dr. Sciarra sent Dr. Lovelace his first case of CMT at the Institute.

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 please call the office at 1-800-606-CMTA.



GENETICS BOOKLET Now Available

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

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

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

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

| | QTY | COST | TOTAL |
|--|-----|--|-------|
| Charcot-Marie-Tooth Disorders: A Handbook for Primary Care Physicians | | active members \$15 inactive members \$20 | |
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VISIT OUR HOMEPAGE

at

www.charcot-marie-tooth.org

GRANTS

(Continued from page 2)

BAYLOR COLLEGE OF MEDICINE GRANT: SEARCHING FOR CAUSES OF CMT

Hiroshi Takashima, MD, PhD, is the recipient of a post-doctoral grant to study new genetic causes of CMT and, thereby, understand the pathophysiology of these disorders. He is working in the laboratory of Dr. James Lupski, a long-time CMT researcher and member of the CMTA's Medical Advisory Board.

Dr. Takashima hopes to recruit patients who have been diagnosed with CMT but have no mutations in the known CMT-associated genes. He will then investigate several new candidate genes, PMP2, MAG, PRX, and CASPR1 to see if they are some of the genes causing peripheral neuropathies, such as CMT.

CMT is a debilitating group of disorders that markedly impair peripheral nerve function. Current therapy does not address the underlying causes of this disorder, but is based on the management of symptoms. This study will focus on identification and elucidation of the molecular defects leading to CMT in order to develop more effective treatments. Further, identification of molecular defects will produce more precise symptomatic, premorbid, and prenatal diagnosis of the various forms of CMT.

COLLABORATION STUDY: HEREDITARY NEURALGIC AMYOTROPHY (HNA)

The final recipient of a CMTA Fellowship is Jan Meuleman, a PhD candidate who is working on a collaborative study between the University of Washington, the University of Antwerp and the University Hospital of Munster, Germany. His sponsor is Dr. Phillip Chance, a member of the CMTA's Medical Advisory Board.

Hereditary neuralgic amyotrophy is an autosomal dominant disease, but it is characterized by recurrent, painful neuropathies, mostly in the brachial plexus, which controls the nerves of the shoulder, arm and hand. Some involvement of the cranial nerves and the diaphragm has been described. Usually an attack begins with severe pain in the shoulder region, lasting for days and followed by muscle weakness and atrophy. Attacks last from weeks to several months and are followed by a slow, but usually complete, recovery. Attacks are often triggered by viral infections or immunizations.

Linkage analysis has located the genetic defect on chromosome 17q25, but the gene is still unknown. It is not known whether the defective gene is expressed in myelin, the axon or one of the tissues surrounding the plexus area, e.g. connective tissue or the immune system. The identification of the disease-causing gene will provide a fast and reliable DNA diagnosis for HNA patients. Since HNA is a recurrent neuropathy with a good prognosis, this research can also yield more insight into mechanisms for the repair of nerve damage.

Members in the News!

Discovery Dateline, a publication of Baylor College of Medicine, recently published an article on a \$500,000 grant awarded by the Vivian L. Smith Foundation. Of that generous gift, \$200,000 will be used to acquire cutting-edge scientific equipment needed to advance research into hereditary diseases of the peripheral nervous system. As the article stated, "James R. Lupski, M.D., Ph.D., Professor of Molecular and Human Genetics, is recognized worldwide for his studies of two of these debilitating conditions: Charcot-Marie-Tooth disease (CMT) and Hereditary Neuropathy with Liability to Pressure Palsies (HNPP). In both CMT and HNPP patients, as well as in patients suffering from many other conditions, nerve cells lose their protective covering, called myelin, which leads to a loss of sensation and fine motor skills. Dr. Lupski's goal is to determine how and why myelin is lost on the peripheral nerves and what intervention might prevent these devastating diseases from being passed on to future generations."

"My colleagues and I are proud to receive the Vivian L. Smith Foundation's outstanding support, which we anticipate will have far-reaching effects," said Arthur L. Beaudet, M.D., Chairman of the Department of Molecular and Human Genetics. "The Smith fellowships will help the brightest young geneticists of today become tomorrow's leaders in molecular medicine, and Dr. Lupski's groundbreaking research on peripheral neuropathies has the potential to yield significant advances for countless people suffering from neurological diseases."

(Dr. Lupski is a member of the CMTA's Medical Advisory Board.)

CMTA Initiates a North American CMT Database

The Charcot-Marie-Tooth Association and the CMT Clinic at Wayne State University, Detroit, Michigan, are in the process of establishing the “CMTA North American Database” to be housed at Indiana University. This database will contain clinical and genetic information on patients with CMT in North America. A generous grant from the Buuck Family Foundation will support the database in its first year.

During the past decade, rapid advances in molecular genetics have generated great excitement in the field of Charcot-Marie-Tooth disorders, both for patients and researchers. The genetic causes of CMT1A, CMT1B, CMT1X, CMT2E, CMT4B and HNPP are now known. Preliminary studies of how mutations in particular genes cause CMT have begun. Animal models of several forms of CMT have been created and are being studied to determine how the disease is caused. However, these advances have not yet led to cures for CMT, or to a thorough understanding of how mutations in particular genes cause peripheral neuropathy.

Progress in four areas must occur for the speed of research to increase in these areas. First, investigators studying genetically known forms of CMT need to have access to larger numbers of uniformly evaluated patients. Research scientists frequently do not actually evaluate patients. They depend upon descriptions of others to determine the disability caused by particular types of CMT. Often, only small numbers of patients are evaluated by any one physician. It thus becomes difficult, if not impossible, to determine the spectrum of disease caused by mutations in a particular gene. Second, investigators trying to identify mutations causing CMT in which a genetic cause is not yet known, such as in CMT2, need access to large numbers of candidate patients. This does not mean that these forms of CMT are always rare. Rather, it means that the investigators don't have access to the patients because, for example, they live far away and don't know about the research. Third, even when new genetic causes of CMT are identified, it is difficult to determine the frequency and range of disability of the new form without evaluating large numbers of candidate patients. Finally, clinical trials in CMT are often limited by the

Rapid advances in molecular genetics have generated great excitement in the field of CMT disorders.

lack of large numbers of carefully evaluated patients to treat.

Confidentiality about patients and their families will be strictly maintained. No patients will be entered into the database without their permission. Data from the database will be made available to all qualified researchers performing research on the cause and treatment of CMT.

Clinical data will be provided by patients, and, with the patients' consent, by their physicians. Information about the database, including how to enroll, will be provided by the CMTA through its web site and newsletter. The data will be entered on specific forms. Athena Diagnostics has generously offered to supply forms to either referring physicians or patients depending on how this can be arranged. Instructions on how to fill out the forms and where to send them will be included with the forms. You will read more about this when the criteria are fully established. When available, forms will be found on the CMTA web site and the web site of the CMT Clinic at Wayne State University. Links to obtain the forms online will be provided by the Muscular Dystrophy Association (MDA) on their web site <http://www.mdaua.org> and by the Neuromuscular Section of the American Academy of Neurology through their newsletter.

Once data is entered in the database, it will be made available to all qualified investigators, upon request. Patient names will be changed to numbers as part of an extensive process to ensure confidentiality and anonymity for patients and their families. Additional information on the registration process will follow in subsequent newsletters.

The CMTA Board of Directors
The CMT Clinic at Wayne State University
The Department of Genetics,
Indiana University

RESEARCH STUDY

Be a participant in an exciting research project! Patients often call the office wishing they were close enough to a research center to participate in some cutting-edge CMT research. Soon, all diagnosed CMT patients in North America will have the opportunity to help researchers by completing the forms or having their doctors do so. The inclusion in the database of vital information about CMT patients will speed research into the understanding of the various forms of CMT and the development of treatments. This project will be for everyone diagnosed with CMT! Watch for further details.



PROFESSIONALS WORKING FOR CMT PATIENTS:

Dr. Steven S. Scherer

(Editor's note: Dr. Scherer is a newly elected member of the Medical Advisory Board and has recently hosted a patient-family conference at the University of Pennsylvania. He is involved in the organization of a Philadelphia-area CMT support group. On the personal side, Dr. Scherer is married and the father of four who lives in suburban Philadelphia with his family. He sees about one new CMT patient each week and has been actively involved in CMT research since 1993.)

Dr. Scherer writes of his medical background and the events which brought him to an active involvement with the CMTA:

“As an undergraduate student at the University of Michigan, I considered careers in both science and medicine. My initial encounters working in a scientific laboratory did not go well, so I planned on going to medical school. Then, I worked as a research technician for a year in the laboratory of Dr. Stephen Easter at the University of Michigan. He was interested in how nerves develop and it was my job to help him conduct the experiments. By doing these experiments and reading, my interest increased in both science in general, and peripheral nerves in particular. My interest solidified into a career when I began my own research project in Dr. Easter's laboratory after my first year of medical school. Thus, I took a leave of absence from medical school to obtain a PhD in order to pursue a study of how peripheral nerves regenerate. As I read the scientific literature, I realized that some of the most important contributions to this field were made by neurologists, such as Drs. Albert Aguayo, Arthur Ashby, Peter Dyck, John Griffin and P. K. Thomas. When I returned to medical school, I was committed to training in neurology, so I could pursue my goal to be a scientific neurologist who specialized in peripheral neuropathy. I was fortunate to be selected by the neurology program at the University of Pennsylvania. Drs. Asbury, Mark Brown, David Pleasure and Austin Sumner were splendid teachers and role models in the diagnosis and treatment of peripheral neuropathies.

“As I was finishing my residency, Dr. John Kamholz (now at Wayne State University)

returned to Penn from his post-doctoral fellowship, in which he had worked on the molecular biology of myelin. Over a cup of coffee in 1988, we discovered we had a mutual interest in peripheral nerve, and we have been friends and collaborators ever since. As a post-doctoral fellow in Dr. Kamholz's laboratory, I learned the molecular approaches that proved to complement the anatomical ones that I had mastered during my previous work. As luck would have it, Dr. Kurt Fischbeck (now the head of Neurogenetics at the NIH) and Dr. Kamholz were interested in CMT, and had adjacent laboratories. I became familiar with the project mapping the CMTX gene which was being carried out in Dr. Fischbeck's lab. Ultimately, we found that only connexin32 was expressed in the peripheral nerve and, since it is a gap junction protein, we considered what role gap junctions play in the myelin sheath. We have recently confirmed that gap junctions

allow small molecules and ions to take a “short cut” across the myelin sheath.

“As you can see, my involvement with CMT research grew out of my interest and my being at the right place at the right time. Since 1993, I have devoted most of my effort to CMT research. As one of the physicians and scientists who are interested in CMT, I realize that patients want treatments that work, but there are many questions that must be answered before a “cure” or even an effective treatment will be found. The CMTA has been both a valuable resource for patients and for scientists who are working on CMT. The CMTA has generously supported research, including a post-doctoral fellow, Dr. Edgardo Arroyo, who works with me. Dr. Arroyo is finding out how myelin sheaths organize the molecules of axons that are needed to conduct electrical impulses. We think that demyelination leads to disorganization of the axon's molecular architecture, which could, in turn, lead to axonal damage. Figuring out why axons are damaged in CMT is one of the promising approaches to treatment.”

Dr. Scherer is the William N. Kelley Associate Professor of Neurology at the University of Pennsylvania. He can be reached through his assistant Tracey Sears at 215-349-5313.



Continuing Our Patient-Education Program: New Orleans Conference Scheduled

The first patient-family conference of the new year will be held at Tulane University School of Medicine in New Orleans, Louisiana, on April 21, 2001. The conference is co-sponsored by the CMTA and Dr. Carlos Garcia, Tulane University School of Medicine, and a member of our Medical Advisory Board.

The meeting will be held in the First Floor Medical School Auditorium and will feature presentations by Dr. Garcia and by Dr. James R. Lupski from the Baylor College of Medicine. Dr. Lupski, CMTA Medical Advisory Board member, is a leading authority on CMT and the researcher who helped discover the chromosome 17 location for CMT1A. Other presenters will discuss such topics as physical therapy, surgery and issues of pain and coping.

The cost for the full-day conference, including a light breakfast and lunch, will be \$40.00 for active members of the CMTA and \$50.00 for non-members. The conference will run from approximately 9 am until 4 pm. An application form will be mailed out to members living in the near vicinity of New Orleans and a form will also be available on the home page at www.charcot-marie-tooth.org

If you are interested in attending and do not live in the area, please call the office and leave your name and address and a form will be mailed to you as soon as they become available.

Other conferences planned for this year include one in Downey, CA, one in Detroit, MI and one in Miami, FL. The conference in Downey will be held in the spring; the other two will be in the fall.



Aquatic Exercise Makes a Splash!

One of the newest ways to update your exercise regime is to jump in a pool and get moving. Aquatic exercise encompasses almost everything except swimming. Shallow-water walking, deep-water running, water weight training and water yoga are a few of the new exercise classes offered at fitness centers, high-school pools and YMCAs today.

You can choose the program you prefer based on what you are interested in working on. Pool exercise can increase cardiovascular fitness, strengthen muscles, enhance flexibility or do all three. To work on cardio fitness,

you might do a brisk power walk across the pool or do step aerobics on a submerged step. Strength training in the pool incorporates bicep curls and tricep extensions using waterproof weights. Water yoga and water ballet incorporate stretches designed to improve flexibility and soothe sore muscles. Stretching in the pool is much easier because the warm temperature of heated pools increases blood flow to the muscles, making them more pliable.

For shallow-water exercise, water fitness shoes are recommended to prevent blisters and to help your feet grip the pool floor. Deep-water exercise calls for flotation vests or belts that keep your body vertical and your head above water. Waterproof dumbbells and foot fins are excellent for weight training and are available at most sporting goods stores.

A study at Baylor College of Medicine found that adults who exercised in water reaped the same benefits as those who exercised on land and aquatic workouts put less stress on the muscles and joints, resulting in fewer injuries.



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CMTA Australia Publishes New Journal

The Charcot-Marie-Tooth Association of Australia has recently published A CMT Journal, Volume 3, which contains articles on CMT written by professionals in the fields of research, health management and welfare. It has information which is specific to Australia with regard to schools and services available there.

However, it also contains some interesting articles on topics of interest to anyone with CMT. They consider surgery for scoliosis, physiotherapy options, the psychological aspects of chronic illness, and the classification of CMT syndromes, among other issues. The journal is available from the CMTAA for \$22.00 Australian, which includes shipping. Contact the organization by email at cmtaa@lycos.com for further ordering information.

We include a portion of an article written by Dr. Garth Nicholson, a member of our Medical Advisory Board, on how CMT can affect the hands.

Dr. Nicholson writes, "Loss of hand strength can be overcome by using thicker handles on instruments and pens. A critical situation will arise if it is not possible to oppose thumb and fingers, and, in some situations, splints are required to hold the thumb



down so that this opposition can be achieved. Sometimes it is possible to transfer a tendon from a strong finger, which is of little use, like the forth or fifth finger, to operate the thumb or to stabilize the thumb.

There are some forms of CMT in which hand involvement is more obvious than in the common form, CMT1A. These forms are CMTX and CMT2D (mapped on chromosome 7). Such patients have thumb drop, which is almost equivalent to foot drop, and these patients cannot lift the thumb up.

Hand involvement in CMT is not uncommon, usually occurs later, is worse in some forms of CMT and needs a referral to an occupational therapist or a hand specialist."

CMTA Remembrances

Your gift to the CMTA can honor a living person or the memory of a friend or loved one. Acknowledgment cards will be mailed by the CMTA on your behalf. Donations are listed in the newsletter and are a wonderful way to keep someone's memory alive or to commemorate happy occasions like birthdays and anniversaries. They also make thoughtful thank you gifts. You can participate in the memorial and honorary gift program of the CMTA by completing the form below and faxing it with your credit card number and signature or mailing it with your check to: CMTA, 2700 Chestnut Parkway, Chester, PA 19013.

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In honor of (person you wish to honor)

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Amount Enclosed: _____

Check Enclosed VISA MasterCard

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Practical Ideas That Help: Products Worth a Try!

SHOES FOR COLD WEATHER

One of our members, John Kathrein, recommends the N.E.O.S. Performance Overshoes for protection from the cold and snow. He has used them for years and they are readily available from a variety of sources. Their homepage claims that the NEOS are better than a boot because they offer the warmth and traction of a winter boot, with the added convenience of allowing you to wear your favorite dress shoes, sneakers or sandals all winter long. The rugged waterproof uppers are lightweight and fold flat for travel, while still protecting feet from the damaging effects of rain, mud, salt and snow.

For more information, visit their homepage at www.overshoe.com. There are a variety of styles to choose from which range in price from around \$60.00 to \$100.00. They can be ordered on-line or from catalogue companies such as Eddie Bauer.



WALKERS AND CANES

Momentum Medical offers a selection of walkers and canes designed to serve two purposes: support while walking and assistance in rising to a standing position from a seated one. One of their products is called the Rising Star Super-Walker. It features a single-button release for easy folding and can be customized to fit users from 5'3" to 6'5". It has two sets of hand grips to

allow the user to rise from and lower easily to a seated position and to walk with comfort and stability once upright. The SuperCane offers much the same advantage with two hand grips for rising and walking. For more information, call 1-800-644-2263.



COMFORTABLE SOCKS

Seamfree Socks are constructed to assure a smooth fit without pressure points. The socks are made from a unique acrylic fabric which wicks moisture from your skin, keeping your feet dry. These socks come in extra wide for patients with edema.



Evergreen Socks feature a thin, light-weight terry interior with a non-restrictive top which stays up without pressuring the skin.



Ellington Socks are the ultimate for warmth and comfort. They are designed with multiple fabrics, acrylic fibers and a terry lining. They also have a non-binding top and a smooth flat-toe seam for extra protection.

For further information about these products, you can call APEX Foot Health Industries, Inc. at 1-800-526-APEX or visit them on the Internet at www.apexfoot.com

SHOES THAT FIT

Ambulator Footwear: Apex Footwear offers a line of shoes called Ambulation Biomechanical Footwear which features a high, wide toe box, soft padded collars and a firm heel counter for rearfoot stability. The various designs all contain 3 removable insoles for unmatched depth and fitting flexibility. These shoes are also available through Apex (see above.)



Turnpike Comfort Footwear, Flushing, NY offers a new shoe style from P.W. Minor and Sons, called Xsensibles. They are extra-depth shoes which accommodate orthotics while offering a more refined look. Turnpike can be reached at (718) 454-5870.



GADGETS FOR THE HOUSE

If you have a weak grasp, the **Good Grips Peeler** is a good kitchen utensil for you. The ergonomic handle is built



up and easy to hold, even when wet. The soft, rubbery plastic handle has flexible ribbing that bends to conform to your hand. There is a whole line of Good Grips products available wherever kitchen gadgets are sold. The peeler retails for



approximately \$7.00. Another offering from Good Grips is the jar opener which works on lids up to 4" in diameter.

Open doors with a **Leveron doorknob handle**. The 4" lever handle makes turning a doorknob easier for weak hands or wrists. The high-impact resin handle fits over your doorknob and tightens with an enclosed wrench. A set of two costs about \$22.95. Available through the Enrichments Catalog of Sammons Preston, 1-800-323-5547.



Another help for those with weak hands or compromised grip is the **Hole-in-One Key Holder**. This 5" key holder allows you to grasp the handle around the outside or with your fingers through the hollow center, providing extra leverage. House keys and most car keys fit into the slots and are held by a single screw. Keys fold into the handle when not in use. Available through the Enrichments Catalog for \$8.00.



■ WHO ARE THE UNINSURED?

According to the US Census Bureau, more than half (54%) of the 44 million Americans without health insurance are young adults aged 18-34. Fifteen percent are children under the age of 18, 17% are between 35 and 44 and 14% are 45 to 64.



Improving Your Chances For Approval When Applying For Social Security Disability Benefits

By Jacques Chambers

There are a lot of myths and misinformation about disability benefits from Social Security and the application process. This is generally undeserved because, while the Social Security Administration (SSA) is an extremely large bureaucracy, the application process is generally straightforward and usually customer friendly.

There are two primary programs for persons with disabilities—Social Security Disability Insurance (SSDI) and Supplemental Security Income (SSI). The former is for workers and their dependents who have paid into the Social Security system through F.I.C.A. payroll taxes, and the latter program is based on need. The process of applying for them is similar.

There is a belief that it is very difficult to get disability benefits from Social Security. I've even heard it said "Social Security always turns you down the first time." Neither is true. Unfortunately, many people apply without any preparation or understanding of the process and that does result in a large number of initial turn-downs, but knowledge of the process can make it easier and increase your chances of approval.

WHAT DOES "DISABLED" MEAN?

First, you need to know what standard Social Security uses to determine if someone is disabled enough to qualify for benefits. Whether it's SSDI or SSI, they define *disability* as: "the inability to engage in any substantial gainful activity by reason of any medically determinable physical or mental impairment(s) which can be expected to result in death or which has lasted or can be expected to last for a continuous period of not less than 12 months."

The key to that legal definition is "the inability to engage in any substantial gainful activity." This means that Social Security is looking at functional problems. Lab numbers, names of diseases, diagnoses are only used to support the fact that you are unable to work in any substantial manner. There must be physical or mental symptoms that prevent working and will last for at least one year.

For people dealing with CMT and other neurological disorders, SSA looks for a condi-

tion, either static or progressive, that produces any type of neurological impairment. This can include weakness, spasticity, lack of coordination, ataxia, tremor, athetosis, or sensory loss. However, according to their regulations, "Documentation of motor dysfunction must include neurologic findings and description of the type of neurologic abnormality." They want to know what the symptoms are and what causes them.

They define *motor dysfunction* as: "persistent disorganization or deficit of motor function for age involving two extremities, which (despite prescribed therapy) interferes with age-appropriate major daily activities and results in disruption of fine and gross movements, or gait or station."

Note that they are not looking for any particular diagnosis. They are looking for motor function problems that interfere with major daily activities. They need to see evidence of actual symptoms that prevent someone from functioning. For a child, that is activities that are age appropriate; for an adult, that usually means ability to perform substantial work, i.e. full-time work.

That doesn't mean that you have to be so disabled you can't do anything except sell pencils on a street corner. Generally, they look to see if you are able to do work that would be suitable for you based on your age, experience, training, and education.

MEDICAL RECORDS

Because they need to see evidence of functional problems, it is important that your medical records be detailed and complete. Social Security looks to your medical records as the primary source of that evidence. Therefore, before you even start the application process, it is important that you sit down with your medical records and look them over carefully. How detailed and complete are they? Are there statements and comments about your condition that support an inability to work? Did the doctor report your statements about problems and list all the symptoms you reported? With doctors being pushed to see more and more patients, sometimes records aren't as complete as they should be to support a disability claim. If a symptom or functional problem is not

reported in the record, then it's going to be difficult to get them to accept it as evidence of disability.

If your records are not complete in listing your symptoms, ask your doctor to write an extensive explanation of your condition, itemizing his observations plus what you have reported to him that wasn't included in your record.

SYMPTOM DIARY

Although it may be too late for the initial application, start a symptom diary immediately. This is simply a journal in which you write down all the symptoms that you have experienced each day. In addition to stating the symptom, describe the severity and its impact on you and your activities. For example: *"So tired after trip to doctor, I had to nap for three hours."* *"I started to clean bathroom but was too tired to continue after cleaning the tub."*

FIND A FRIENDLY OFFICE

Social Security offices develop their own personalities. Some are more difficult to deal with than others. This is a good time to put your grapevine to work for you. Talk to others from your support group or your doctor's office or others who have gone through the procedure. You can apply for Social Security Disability at any of their field or branch offices. When you call the national number to make the appointment (800-772-1213) you can request which office you want the appointment made with. If you can find an office that has a reputation of being easier to deal with, go there. It really will improve your chances for approval.

GO TO THE APPOINTMENT PREPARED

When you make your appointment to apply for Social Security, you will be given the option of applying by phone after they send you some forms to complete or of going into the office and applying in person. I recommend going into the office for two reasons. The Claim Representative must report in writing his or her impression of you and your functional abilities, so if you have trouble walking or thinking clearly, the representative will observe and report it. Second, you are asked to provide original documents, such as your birth certificate. If you deliver it in person, they can see the original, photocopy it, and return it to you immediately so you don't risk losing valuable documents in the mail.

Take all the documents you are instructed to take. Normally this will include:

- Proof of birth. If you were born in this country, they want to see an original or a certified copy of your birth certificate. (If you don't have one, go to the appointment; they will help you obtain one.)
- Military discharge papers, if any.
- Social Security card, or at least your Social Security number.
- Proof of residency if a non-citizen.
- Bank records, housing documents, deeds, leases, and other financial documents showing your assets and income (if you're applying for SSI).

There are other documents you can take which, although not required, will help speed up the process:

- Your Medical Records. Although they can get them from your doctors, it will speed up the process if you obtain copies of all your records and take them with you to the appointment.
- Your symptom diary if you've been keeping one.
- A letter from your doctor detailing your condition and presenting his or her observations about your ability to function at a job.
- Third-party testimony. These are letters from people you live with or work with that detail their observations of your deteriorating functional ability. Anecdotes about problems they have observed can be very helpful. A statement from a co-worker or supervisor about increasing difficulties in performing your job can be especially persuasive.

THE CONSULTATIVE EXAM

Occasionally an applicant will receive a letter announcing they have an appointment with a physician for a consultative exam. If you receive one, immediately call your Claim Representative, the name and number will be on the letter, and ask that your own doctor perform the exam instead. Consultative exams are notoriously brief and superficial and rarely provide support for a claim. Social Security's own regulations give preference to the attending physician's information, so ask for a supervisor if you have trouble getting them to agree to using your own physician.

(continued on page 15)

Coping with CMT: Making the Choice to Be a Survivor

By **D. R. Schwertfeger**

(Editor's note: Mr. Schwertfeger has a B.A. in psychology and a Masters in sociology. He has written for the newsletter in the past on orthotics, exercise, and "being a hero.")

Every day, we face difficult situations, problems and crises, and we have to cope. Life's challenges come in all shapes and sizes, so there's no one way of coping with everything, but we do tend to fall into general patterns of responding to problems. Some of us, for example, always confront things head-on. We take charge and aggressively pursue solutions so that we can get matters resolved and get on with life. Some of us procrastinate and avoid doing anything until we absolutely have to. In the meantime, we hope someone will help us out or that things will work themselves out and just go away. And some of us refuse to cope. We deny that problems even exist and simply refuse to deal with them.

Learning to cope with CMT or any of life's disappointments is how we make life worth living. It will be easier for some and harder for others, for our success will depend on the understanding and acceptance of those we love, our families and friends, as well as the path we have chosen to take in life. What's important is that we can *choose* how we cope with CMT. Greatly oversimplified, the choice we make will determine whether we're victims or survivors of CMT.

I chose to be a survivor, but it wasn't a conscious decision, not at first anyway. I went through a few phases first, like the "oh-my-God-my-life-is-over" phase. When I was first diagnosed with CMT and was told about its being progressive, the only questions I had for the doctor were: a) how bad is my impairment going to be? and b) how soon is it going to happen?

He couldn't give me a definitive answer to either one, of course, but what he did tell me

was scary enough. Until then, I had taken my physical well-being for granted. I played baseball and football. I went hiking and hunting. I climbed mountains and I spelunked. I was just 18 years old and I was suddenly contemplating the end of my life as I knew it.

That was pretty overwhelming, but over the past 25 years, I've come to realize that the progression of CMT is a lot like the process of aging. We all know that some day we're going to grow old and gradually lose certain abilities. We know that, but we don't spend our youth dwelling on thoughts of what we'll look like and what we'll no longer be able to do when we're 70. We just deal with, and accept, the changes as they happen. That's how I've dealt with the progression of my CMT—one day at a time.

The second phase I had to work through had to do with the discovery of my identity. As a teenager, I was trying to figure out what I wanted to do with my life. Learning I had CMT narrowed my career choices somewhat, but I've never allowed my being a CMT patient to become my identity. To do so would have placed limitations on every aspect of my life. I'm not a patient; I'm a writer, a husband, a father and I do everything I can to enjoy, and make the most of, my life.

If that sounds a little like I'm in denial of my CMT, I won't completely disagree, but it's a healthy state of denial that's helped me through the why-me? phase. I am denying myself the opportunity to wallow in self-pity. There have been times when I've lain awake wondering, "Why me?" Why can't I run and jump and wiggle my toes like the millions of people who take those abilities for granted, just as I did before CMT began ravaging my body. I also confess to having an occasional self-indulgent moment when I want to give in and say "I can't" rather than, "With a little help, I think I can," or when I want people to look at me and say, "Poor fellow, he's got CMT."

But, unavoidable as those feelings sometimes are, I try not to dwell on the unfairness of



Mr. Schwertfeger believes in "dealing with the changes of CMT as they happen."

the hand fate has dealt me. There's nothing I can do about it and I have yet to accomplish anything by feeling sorry for myself or asking others to feel sorry for me. Self-pity just isn't productive, so rather than being a "poor fellow with CMT," I'd much rather be regarded as someone who accomplishes a lot in spite of having CMT.

I didn't get through that phase overnight. In fact, it's taken me years to come to grips with the emotional consequences of my physical limitations. Although I may not see myself as a CMT patient, that doesn't mean that CMT hasn't had an impact on me. In a way, I consider myself fortunate to be in a profession that draws its esteem from intelligence and craft, not sheer masculinity, but that doesn't keep me from having feelings of inadequacy about being unable to do "manly" things like taking my son hunting or playing football with him.

It's also been difficult learning to accept help, and even more difficult to request it. Even now, each time the progression of my CMT causes me to lose the ability to do something,

I devise elaborate strategies to disguise the fact that I can't do it.

Of course, it's not always obvious that I have CMT until I have to say that I can't do something. Once unmasked, however, I've grown more comfortable with allowing other people to do things for me, and I find it easier to make my excuses when I'm unable to help with tasks like carrying heavy boxes or moving pieces of furniture. I'm stubborn and self-reliant, but I'm not so foolhardy as to injure myself or someone else. I'll let someone carry the heavy load and I'll assist in whatever way I can.

What I didn't realize until my wife and I began discussing all these issues was how much talking helped both of us. By not complaining when something bothered me, I had thought I was sparing her from dealing with my problems. Little did I know, however, that she felt shut out and frustrated that she was unable to help. With her love and understanding, I'm able to take charge and confront the problems CMT presents, and we're surviving just fine.

DISABILITY BENEFITS

(Continued from page 13)

SPEND TIME COMPLETING THE FORMS

When you first apply for Social Security disability, you will be asked to complete some initial forms. This is simply information about yourself, names and addresses of your medical providers so they can get your records, and a history of all the types of work you've done in the past fifteen years. If you're applying for SSI, there will also be forms to complete concerning your financial condition, what you own, what income you have, etc.

After a couple of weeks, you will receive more forms to complete, these dealing more specifically with your condition and its impact on your life. For people dealing with CMT symptoms these questionnaires are likely to concern any fatigue and pain you may be experiencing. They will also send you forms asking about your daily activities and how you accomplish the routine tasks of daily living. There may be other questionnaires as well, based on what they find or don't find in your medical records.

You should spend some time and fill these forms out carefully and completely. Don't leave any blanks; write "N/A" if it doesn't apply to you. When filling them out, keep in mind your bad days—not the days when you're feeling fine.

It is best if you fill these forms out yourself. The Analyst reviewing the form will look at how you filled it out as well as what you say. Don't worry if you have to make corrections or changes. Just line them out and re-write it. You won't be graded on neatness, but make sure you print clearly enough so that it can be read. Use blue or black ink when completing the forms. You can also put information on additional sheets of paper if there's not enough room on the form, but be sure to clearly label the answer and make sure your name and Social Security number is on each sheet.

If you use a computer or typewriter or have someone fill it out for you because you are unable to write legibly, explain that on the form and tell why. If the form was especially difficult for you to complete, also explain that and include how long it took you to fill it out. Everything will be considered when reviewing your claim.

No one can guarantee that your claim for benefits will be approved, but with preparation, thorough and complete medical records, and a focus on functional symptoms, your claim will have a better chance of being approved.

Jacques Chambers is an independent benefits counselor. He spent twenty-five years in the health insurance industry and the last ten years assisting people with their public and private benefits. He was diagnosed with CMT in 1994. He can be reached at jacquesmc@earthlink.net

CMTA Support Group News

■ Arkansas - Northwest Area

The Northwest Arkansas Area Charcot-Marie-Tooth Support group last met on November 18, 2000. We meet at the Jones Center every third Saturday. Our next meeting, scheduled for December 16, 2000, was canceled due to ice and snow and dangerous traveling conditions. Those with CMT should not take the risk of fall or injury in weather such as we've had the last few weeks.

We were very encouraged by the last meeting attendance. It was great! We had 13 in attendance, including a few new members and some who hadn't been with us in a while. Our top attendance has been 22 in the spring. Our group has recently joined with the local peripheral neuropathy group. Their group has had low attendance, so three of their members, including their leader, have joined our group.

We continue to feel that our efforts are paying off and we are reaching more people all the time. We grow by one or two every month. To date, we have approximately 59 on our list, up from three at our first meeting in November of 1998. It is very encouraging.

One of our members who works with hospice has agreed to do one of our meetings on "Grief." At first glance, this might not seem to be a logical topic for us, but the process of "loss of function" is very similar to the grieving process you go through when you lose a loved one. We look forward to having her help us with the

process of denial and then acceptance in dealing with a chronic illness.

The group has changed its meeting times to accommodate those who travel a distance. Now the group meets in the Jones Center for Families in Room 206 from 11 am to 1 pm. This is a new room with better accessibility. Readers can access information about their support group by visiting www.geocities.com/charcot-marie-tooth

Libby also has a new email address and telephone number. Please see the contact list for that information.

■ California - Berkeley Area

The November meeting featured a lively interchange of shared ideas. One member gave information about the possible benefits to nerve health of methylcobalamin, a more easily absorbed form of vitamin B-12. Another member suggested wearing the rubber finger caps that bank tellers use to help pick up small or slippery objects when numb fingers are a problem. Another showed a "ring pen" which slips over the index finger and makes writing easier and one younger member discussed the "Dr. Grip" pen, which is easy to hold and use.

The next meeting on January 20, 2001, featured Dr. Gail Widener, a physiologist, who discussed physical therapy and exercise for people with neuromuscular diseases. Members were encouraged to ask questions.



New Support Group Forming!

The first meeting of the Philadelphia area support group will be on Saturday, March 24, 2001, from 9 am to 11 am at the Hospital of the University of Pennsylvania. The meeting will be held on the third floor of Founders Building, directly above the cafeteria, in Plaza room A. Dr. Steven Scherer, Neurologist, will be in attendance at the first meeting.

The support group organizer is Amanda Young, who can be reached at 215-222-6513 or by email at stary1@bellatlantic.net

The impetus to get this group going came from the attendees at the October patient/family conference at the University of Pennsylvania, who were uniformly interested in having a support group in this area.

Amanda Young attended the Human Genetics Meetings in Philadelphia as a representative of the CMTA.

CMTA Support Groups

Alabama/Greater Tennessee Valley

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

Arkansas—Northwest Area

Place: Jones Center for Families,
Rm. 206
Meeting: 3rd Saturday of each month
Contact: Libby Bond, 501-795-2240
E-mail: charnicoma57@yahoo.com

California—Berkeley Area

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

California—Los Angeles Area

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

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

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

Colorado—Denver Area

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

Florida—Boca Raton to Melbourne

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

Florida—Miami/Ft. Lauderdale

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

Kentucky/Southern Indiana/ Southern Ohio

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

Massachusetts—Boston Area

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

Michigan—Detroit Area

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

Michigan—Flint

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

Minnesota—Benson

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

Mississippi/Louisiana

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

Missouri/Eastern Kansas

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

Missouri—St. Louis Area

Place: St. Louis University Medical
Health Center
Meeting: Quarterly
Contact: Carole Haislip,
314-644-1664
E-mail: c.haislip@att.net

New York—New York City

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

New York—Horseheads

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

New York (Westchester County)/ Connecticut (Fairfield)

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

North Carolina—Archdale/Triad

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

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

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

Ohio—Greenville

Place: Church of the Brethren
Meeting: Fourth Thursday,
April–October
Contact: Dot Cain, 937-548-3963
E-mail: bobcain@wesnet.com

Oregon—Willamette Valley/ Pacific NW

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

Pennsylvania—Philadelphia Area

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



WRITE TO US!

Pat Dreibelbis, Editor

The CMTA Report

CMTA

2700 Chestnut Pkwy.

Chester, PA 19013

The CMTA reserves the right to edit letters for space.

Ask the Doctor

Dear Doctor,

Does TES (threshold electrical stimulation) help with Charcot-Marie-Tooth disease (CMT) at all? I have heard anecdotal reports that TES has helped two children in California to get out of AFOs, improve their balance, and even put muscle mass on their lower calves. Do you have medical evidence that this procedure does, indeed, work for CMT patients?

A CMTA Medical Advisory Board member replies,

I am not personally aware of any medical evidence that TES is helpful in treatment of CMT and am skeptical of stories about patients no longer needing AFOs or orthotic support due to TES. It is our feeling that good rehabilitation medicine, and, when appropriate, an exercise program, are likely to be more productive approaches to therapy.

Dear Doctor,

Our daughter, who is 23, was diagnosed with Charcot-Marie-Tooth disease nine years ago. She has undergone surgeries for heel cord lengthening and scoliosis.

I've heard that CMT patients should avoid alcohol consumption. Would you please elaborate with an explanation of the dangers of alcohol? Can CMT patients drink socially or occasionally?

An MDA Clinic Director Replies,

Alcohol can damage muscle and nerve. Daily consumption of alcoholic beverages or episodic consumption of significant amounts of alcoholic beverages is likely to damage muscle and nerve. There is a condition known as alcoholic neuropathy. A person who already has a disorder impairing the peripheral nerves

Letters to the Editor:

Dear CMTA,

I was scheduled for toe surgery. Was I worried about the skills and qualifications of my doctor or the other people who would be caring for me? No, I was worried about being cold before, during and after the surgery. I have CMT. Being cold in my feet used to be my problem, but they don't seem to feel the cold so much anymore. I'm not sure that is a good thing, but my legs, especially in the front, and my arms suffer from being cold, sometimes even in quite warm weather. Not only do they feel cold, but if I don't take some measures to warm them, I start to get weak and can literally feel like I'm being destroyed physically.

Back to the day of the surgery, December 27th, in Minnesota. You get the picture. I wore long underwear and warm slacks and we even parked in the handicapped parking right by the door. My legs were still cold. After a brief wait, I was told to go into a room. I told the woman in charge that I suffer from the cold, but apparently she didn't quite understand because she told me to put on two thin garments she gave me (which, of course, did not even cover my legs

and arms) and some little booty-like socks. I stared at the clothes and I wanted to cry. I couldn't put them on. I told my husband that I didn't think she had understood what I had told her, so he went to talk to her in the hallway. I could hear him explaining to her what the cold does to me and the fact that I had not put on the thin dressing gowns.

She told him she had not realized how severe the problem was and she would take care of it. She told me to put on the gowns and come out quickly and she would see to it that I would be warm. (Why do I take this disease so personally, as though it were my fault, and therefore, have a hard time asking for the things I need?)

I did as instructed and she led me down the hall and onto a bed. Then, the most heavenly thing happened. They put a plastic sheet on me which was hooked to a machine that pumped warm air into it. Then, they put a thin blanket over me. It felt so good. I don't remember the surgery room, but I'm sure I had this on me the whole time because it was on me when they wheeled me down to surgery and it was still on me when I woke up after surgery. They told me it was called a "Bear Hugger."

needs to avoid doing anything that would make the situation worse. It is unlikely that drinking an occasional beer, glass of wine, or mixed drink will cause a problem. With alcohol, like other chemicals, “the dose makes the poison.”



Dear Doctor,

Over the years, I have tried a variety of medications to treat my CMT pain. Recently, I was advised to take Neurontin (brand name for gabapentin). A whole new pain-free life has been opened up for me. I've tried to find information about how Neurontin works to stop the pain, and give me stamina and better balance. Have there been any studies on the benefits of Neurontin for people with CMT?

I want to thank the CMTA for the doctor I found (on their list for Minnesota), the letter that explains the disease and the list of neurotoxic drugs. My anaesthesiologist praised the letter and the list. I am also blessed with a very supportive spouse.

If anyone has questions about the “Bear Hugger” I can be reached at 320-567-2156 or email at rrmills@info-link.net.

—Rosemary Mills, MN



Dear CMTA,

I wanted to drop you a short note to tell you how much your information and help on the phone has meant to my family. My daughter was diagnosed with CMT only a few months ago and I was feeling very isolated and fearful. I was lucky that the pediatric neurologist who diagnosed her gave me your office number. When I called, I was given really helpful information about contacting Shriners and the MDA and a support group in our area. I have done all of those things and am feeling much more in control of our situation now. I also received your helpful packet of informa-

A CMTA Medical Advisory Board member replies,

Neurontin is one of a group of medications that are used to treat painful peripheral neuropathies. As with the other medications utilized for this “neuropathic pain,” including amitriptyline, nortriptyline and carbamazepine, Neurontin is not specific for CMT pain but is used in many painful neuropathies. Unfortunately, none of these medications works all the time for all patients; some work for some patients, others work for other patients. It is not easy to predict which medication will work for which patient in advance. While there are thoughts on how some of these medications work, how Neurontin stops pain is not clear. Of interest is the fact that other anti-seizure medications, like phenytoin (Dilantin) or carbamazepine (Tegretol), also treat pain in some patients with neuropathy.

tion and was impressed with how quickly it came after I called you. Your newsletter has so many helpful articles, especially about braces and social security issues.

So, thank you for being there. The upbeat attitude of the person I spoke with was just what I needed in facing the unknown future of my daughter. It's really wonderful to know that there is someone I can call and just talk to who understands what I am going through. Since no one else in our family has any signs of CMT, it's difficult to foresee what might develop for Karen and it's very reassuring to know that there is help for us in this difficult time.

—P.S., CA



Dear CMTA,

My son David was born on October 26, 2000. I was able to have the blood from the umbilical cord tested to determine if David has CMT or not. We are fortunate that he tested negative. Please let others know that they can have the blood drawn at birth and the sample sent to Athena Diagnostics for testing.

—L.G.

The CMTA Report

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MEDICAL ALERT:

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

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

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

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



What is CMT?

- ... is the most common inherited neuropathy, affecting approximately 150,000 Americans.
- ... may become worse if certain neurotoxic drugs are taken.
- ... can vary greatly in severity, even within the same family.
- ... can, in rare instances, cause severe disability.
- ... is also known as peroneal muscular atrophy and hereditary motor sensory neuropathy.
- ... is slowly progressive, causing deterioration of peripheral nerves that control sensory information and muscle function of the foot/lower leg and hand/forearm.
- ... causes degeneration of peroneal muscles (located on the front of the leg below the knee).
- ... causes foot-drop walking gait, foot bone abnormalities, high arches and hammer toes, problems with balance, problems with hand function, occasional lower leg and forearm muscle cramping, loss of some normal reflexes, and scoliosis (curvature of the spine).
- ... does not affect life expectancy.
- ... has no effective treatment, although physical therapy, occupational therapy and moderate physical activity are beneficial.
- ... is sometimes surgically treated.
- ... is usually inherited in an autosomal dominant pattern, which means if one parent has CMT, there is a 50% chance of passing it on to each child.
- ... Types 1A, 1B, 1X, HNPP and EGR-2 can now be diagnosed by a blood test.
- ... is the focus of significant genetic research, bringing us closer to answering the CMT enigma.

The CMTA Report

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



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