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The CMTA Report

OCTOBER 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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Occupational Therapy and CMT

By **SUSAN SALZBERG, OT**

For people with CMT, the occupational therapist (OT) is a medical professional worth getting to know. I may be a little prejudiced in this opinion because I am an OT. I got my degree and sat for my licensing exam in 1971 and have worked steadily ever since. In 1976, following the birth of my son, I was diagnosed with CMT.

In one sense it was a shock to be diagnosed with CMT, considered at that time to be a rare condition. I had thought my "klutziness" was a stable condition and was not prepared for the progression I had during pregnancy. Finding out I had a genetic condition just as my son was born was also a blow. But in another sense it was a relief to acknowledge that I had a real problem and did not just "walk funny" because I wasn't trying hard enough. There was nothing that could prepare me for being a Mom, but my training as an OT helped me cope with my new diagnosis.

An OT's education has a broad base. The courses an OT takes can be grouped into three general categories:

- 1) How The Body Works: human development, anatomy, physiology, and general medical conditions,
- 2) How The Body Thinks: (psycho-social issues): coping mechanisms, general and abnormal psychology, and dementia intervention, and
- 3) What To Do When The Body Doesn't Work: (rehabilitation issues): alternative methods of performing self-care, energy conservation and work simplification, adapting the environment, and hand therapy. Like psychologists, social workers, physical therapists, or teachers, OTs are employed in diverse environments. Most common job settings for an OT are school systems, settings where phys-



Susan Salzberg demonstrates the dynamometer, which measures grip strength.

ical rehabilitation is done (hospitals, nursing homes, hand therapy clinics) and psychiatric hospitals.

A person with CMT most commonly would consult an OT for problems related to:

- Poor balance—difficulty managing self-care tasks that require standing.
- Hand Problems—hands can be weak, have reduced sensation and are thus "clumsy."
- Fatigue—inability to complete tasks that need to be done.
- Developmental Problems—since the above problems can affect development, children may receive OT (and/or PT) services in the school from a pediatric OT.

I will describe three scenarios of three people with CMT (names changed, of course) who have received OT treatment.

JAMES

James is a 67-year-old whose CMT was diagnosed 35 years ago while he was in the military. He retired with a medical disability and receives his care from the VA Hospital where I work. James has taken advantage of the VA's generous policy of providing all medically necessary equipment to qualifying disabled veterans.

(continued on page 4)

Hope for the Future

By **VINCENT BERTOLINO, Executive Director**

The CMTA.... Living our Mission

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

Vision Statement:
Dedicated to
generating hope.

I am writing this article on the day terrorists attacked the World Trade Center, reducing it to rubble. Such a tragedy makes it difficult to write about "hope for the future." My condolences go out to the families who have suffered a loss due to this terrible act. Those of us who live in the aftermath must continue to live, and endeavor to make the world a better place. Whatever the future may hold for us, we must keep our vision of what the future should be and make plans for actualizing that future.

Over the past five weeks, we have made some significant changes that will enable the CMTA to better serve you and provide more funds for CMT research. Pat Dreibelbis will now be in charge of all CMTA literature as our Editor in Chief. Edna Rowdon will be the CMTA's Accounting Supervisor. Dotty Lilley will be Program Coordinator, overseeing the scheduling and management of meetings and events.

The website has been revised and improved. Thank you for your feedback; we have endeavored to provide the features you have requested. Please visit our website and feel free to let us know what you think. If you shop on the web, visit the CMTA's shopping link. You can do your holiday shopping there at many national brand retailers who have pledged a portion of the sale to support our cause.

The CMTA assisted the CMT Clinic at Wayne State in raising nearly \$45,000 for funding operations at that institution. Dr. Michael Shy and his staff see hundreds of patients with CMT

and this funding will enable his group to continue to provide these services.

Mr. Patrick Torchia organized the First Annual Johnstown, Pennsylvania CMT Golf Outing, raising nearly \$20,000 for the CMTA. Thank you Mr. Torchia!

The First Annual John J. Scarduzio Golf Outing raised more than \$30,000. This is an outstanding beginning for a new CMTA fund-raising tradition. A warm thanks and applause to Chris Scarduzio for establishing this event.

We have also begun to aggressively pursue corporate funding for a number of research projects in the pipeline. This isn't just a matter of calling up and asking for money. We must start with an understanding of what is needed, then define the projects, their costs, aims, and benefits up front. Then we identify organizations within these areas of interest that might be willing to partner with the CMTA to make these programs a reality.

Last but not least, the CMTA is explicitly defining its role as an active participant in leading the way to a cure. We will have two medical advisory board meetings this year, involving neurologists and geneticists. Providing a forum for greater discussion about the future directions of CMT research is a critical part of this leadership.

The number of genes that cause CMT suggests that there are many mechanisms of disease, each with its own therapy or cure. Consequently, the research task ahead will not be easy, but, if we break it up into a series of manageable projects, funded by many individuals and organizations, we can make this happen.

Keep the vision of the cure, and thank you for your continued support!

UNIVERSITY OF WASHINGTON NEUROLOGIST TO GIVE LECTURE ON NEUROGENETICS ETHICS

Thomas Bird, MD, a member of the CMTA's Medical Advisory Board, will receive the George W. Jacoby Award from the American Neurological Association and will present the Jacoby Lecture on Oct. 1 at the Association's 126th Annual Meeting. Bird is a professor of neurology and medicine, chief of the Division of Neurogenetics in the Department of Neurology and serves on the research faculty in geriatrics at the Veterans Affairs Puget Sound Health Care System. He also collaborates with UC Davis School of Medicine clinical professors on the research project, "Risks and Benefits of Genetic Testing in Persons with Hereditary Neuromuscular Disease," funded by the NIDRR Rehabilitation Research and Training Center in Neuromuscular Diseases, Dept. of Physical Medicine and Rehabilitation. The Jacoby Award is presented every third year to an American Neurological Association member who has done especially meritorious experimental work. Dr. Bird will be honored for his study of hereditary neurological disorders including ataxias, Charcot-Marie-Tooth neuropathy, Huntington's disease and spastic paraplegia. At the Jacoby Lecture, Dr. Bird will discuss ethical issues in neurogenetics that are emerging from genomic neurology and that will confront most neurologists in the near future. Dr. Bird can be reached at 206-764-2308 or tomnroz@u.washington.edu

News of his research may be found at the following web sites:

<http://www.rehabinfo.net/RRTC/Newsletters/ProgramOverview99/GeneticTesting.html>

<http://www.geneclinics.org/profiles/cmt/>

<http://depts.washington.edu/neurogen/>

Wayne State Hosts First Annual Charity Golf Tournament

On July 23, 2001 Wayne State University hosted their first annual charity golf tournament at Barton Hills Country Club in Ann Arbor, Michigan. Wayne State is the site of the first comprehensive CMT clinic in the US.

The charity golf tournament was a tremendous success. The tournament was supported by 131 golfers and over 170 guests for dinner. The weather cooperated and the competition was fierce. The tournament featured thirty-three teams playing a scramble format. There was a buffet luncheon prior to the tournament at Barton Hills. Following the tournament, there was a dinner, raffle and award presentation, which included a brief presentation by Dr. Michael Shy, Director of the CMT Clinic at Wayne State University.

The tournament received a broad base of support from CMT patients and their family and friends from the greater Detroit area. Many healthcare providers supported the tournament by sponsoring holes and other tournament functions. There were also a number out of town guests, including Ann Beyer, Chairman of CMTA and her husband, Ron.

Contributions are still being received, but the tournament is projected to raise in excess of \$40,000 for CMT research. The funds will be used to support patient care and research performed at the Wayne State University CMT Clinic in Detroit, Michigan.

The charity golf tournament was fun for all who participated, but, more importantly, the money raised will provide additional funding for CMT. Plans are already in place for next year's charity golf tournament, to be held in July 2002.



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|---|-----|--|-------|
| Charcot-Marie-Tooth Disorders: A Handbook for Primary Care Physicians | | active members \$15 inactive members \$20 | |
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| CMT Facts II <input type="checkbox"/> English <input type="checkbox"/> Spanish | | active members \$5 inactive members \$7 | |
| CMT Facts III | | active members \$5 inactive members \$7 | |
| CMT Facts IV | | active members \$8 inactive members \$10 | |
| A Guide About Genetics for the CMT Patient <i>No shipping and handling on this item only.</i> | | active members \$4 inactive members \$5 | |
| NEW! Tote Bag | | \$10 | |
| CMT Informational Brochure <input type="checkbox"/> English <input type="checkbox"/> Spanish | | FREE | |
| Physician Referral List: States: _____ | | FREE | |
| Letter to Medical Professional with Drug List | | FREE | |
| Contribution to CMT Research <i>10% will be applied to administrative expenses.</i> | | | |
| Shipping & Handling <i>Orders under \$10 add \$1.50, orders \$10 and over add \$3.00</i> | | | |
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OCCUPATIONAL THERAPY

(Continued from page 1)

I first saw James 10 years ago. He had just been fitted for AFOs and was pleased with his gait and the added stamina he had on his feet. He was in the process of remodeling his bathroom and asked about universal design. Often OTs give pointers on making sure remodeling projects make the home truly accessible and has the modifications the person might need in the future. Since we have a Department of Universal Design as part of the School of Architecture at NC State University in Raleigh, I encouraged James to discuss his remodeling project with them. His home now has a roll-in shower, wide doors with levers instead of knobs, and a ramp.

At some point about 8 years ago James came in for an ADL (activities of daily living) evaluation. He was having difficulty with his fine motor coordination. His grip strength was around 35 pounds (normal is 60-80) and he had some loss of sensation. These are the suggestions I made: 1) Larger clothing is easier to put on and take off. 2) Clothes without fasteners (sweats) are easier to manage. 3) Large handles are easier to grip. 4) Using large muscles (i.e. arms) puts less stress on the joints than using small muscles (i.e. fingers). With those suggestions in mind, James chose the following adapted devices to try out at home: 1) A button aid because sometimes he needs to wear shirts with buttons. 2) The button aid has a large handle and a hook on the other end to help him pull up his zipper. 3) Small plastic loops that fit through his pant zippers to allow for easier

grasp. 4) An under-counter jar opener which allows you to hold the jar in two hands, using the muscles of the arm to turn it to take the top off. 5) Eating utensils, and a vegetable peeler with large handles similar to the Good Grips® line. 6) A kitchen knife with an ergonomic right-angle handle. 7) An electric toothbrush with a handle that is easy for him to grasp (Braun ultra plaque remover®).

James was satisfied with the above items and still uses them. He ambulates in his home, but several years ago started using an electric scooter in the community. One of the OTs in my department evaluates people for electric mobility. In some settings a PT will do this. Recommendations are based on the following: 1) The size of the person. 2) Where the person will use the device (inside or outside or both). 3) How the person will transport the device. 4) Any special modifications such as foot support, back support, and hand use for the driving controls.

DIANE

Diane is a 45-year-old single mother with a teenage daughter. She is employed as a home health nurse, which means a lot of driving and a lot of time spent in her patients' homes. Her CMT got rapidly worse about 5 years ago, with a decline in standing balance and an increase in fatigue. At that time she began wearing AFOs and started using hand controls on her car. She came to see me because of fatigue and problems with hand use (decreased strength, decreased coordination and pain in her thumb joints).

Diane and I addressed the topic of energy conservation and work simplification (EC/WS).

O.T. STRESSES ENERGY CONSERVATION AND WORK SIMPLIFICATION



The under-counter jar opener allows a person to use two hands to unscrew lids.



The button aid helps fasten and unfasten small buttons and zippers.



Good Grips® utensils are all designed with large soft handles.

The goal was to make Diane her own efficiency expert, so we dissected her day and put chores into several categories. A: must do, B: enjoy doing and C: can be done by someone else. It is Diane's choice how she structures her day, but we recommended pacing. This strategy involves alternating a lightweight activity (TV, patient notes) with a heavyweight activity (preparing a meal, doing laundry). We recruited her daughter for several of the "C" chores. EC/WS can involve using adapted devices or modified strategies. People without handicaps use adapted devices. The TV remote control and garage door opener are two common examples. Do you think there will be a time when we all use voice-activated word processors for our computers? This software has become much more financially accessible in recent years and allowed Diane to consider writing patient notes a lightweight chore. She is using many of the same devices as James (button aid, large handle utensils, jar opener and right-angle kitchen knife). Diane opted for using bathroom safety equipment rather than remodeling her home. Her stall shower accommodated a shower chair and we put a clamp-on elevated toilet seat on her commode. She later decided that the elevated toilet seat was not helpful and returned it to the VA for reissue.

It is essential to try out items before purchasing them, because not everything you see in a catalogue or view in a store will be something you can use. The VA Hospital where I work makes this easy. Many occupational therapy departments have sample items on hand to show people. Often hospitals charge a mark-up for stocking the item, and catalogues usually charge a shipping fee. Shop carefully!

Diane also had hand issues but was not ready to consider surgery. Her hands hurt from writing patient notes (a task we eliminated) and were painful when she gripped the car steering wheel. The pain was at the base of her thumb, where there are two joints connecting the thumb to the hand. If the pain is chronic and severe, an OT can construct a thermoplastic splint to support the thumb joints and allow the thumb to be positioned so it can touch each fingertip. (This is called "opposition.") There are also commercially available orthotics which support the same joints. Because Diane could oppose her thumb, and her joints didn't have any bunion-type bony protuberances, she chose two orthotics from our shelf: 1) a Comfort Cool™ Thumb CMC Restriction Splint (available from North Coast Medical, Inc.) and 2) the Collum™ thumb protector. The Comfort Cool™ is a perforated neoprene support lined with terry cloth. In spite of the name, the skin underneath can get sweaty. It wraps around a good portion of the hand, wrist and thumb and provides a neutral warmth, which some people find comfortable. The Collum™ thumb protector, made of leather with a metal stay, provides a surprising amount of support for small surface area of contact. Like a nice leather shoe, it becomes more comfortable as it is worn and absorbs perspiration, making it feel much cooler than any other thumb support. Diane wears the Comfort Cool™ at night if she's had a hard day and her thumbs hurt. She wears the Collum™ during the day. Neither orthotic restricts the use of her hands.

(continued on page 6)



The quad spoon helps the person who has hand weakness.



A zipper pull fits most zippers and provides a larger hand hold for pulling.



The Key Turner adapts vehicle and door keys to have larger bases for holding.

SOURCES

The following mail-order companies are the ones we use most often. There are many others. Most have separate catalogues for “patient-direct” vs catalogues for therapists because some items (like orthotics) should be evaluated and fit by a therapist.

AliMed

297 High Street
Dedham, MA 02026
1-800-225-2610
www.alimed.com

Maddak, Inc.

(Ableware Independent Living)
6 Industrial Road
Pequannock, NJ 07440
1-800-443-4926
custservice@maddak.com

Mercer County Rehab Supply

202 North Broad Street
Grove City, PA 16127
1-800-258-4212
www.mercercountyrehab.com

North Coast Medical Company

18305 Sutter Blvd.
Morgan Hill, CA 95037
1-800-821-9319
www.beabletodo.com
www.ncmedical.com

Sammons Preston

PO Box 5071
Bolingbrook, IL 60440
1-800-323-5547
www.sammonspreston.com

Smith & Nephew Rehab Division

One Quality Drive
PO Box 1005
Germantown, WI 53022
1-800-558-8633
www.smith-nephew.com/
us/rehab

OCCUPATIONAL THERAPY

(Continued from page 5)

VICKI

Vicki, like me, is an OT with CMT. Her specialty is pediatrics, but since she recently had hand surgery she knows much more about what hand therapists do for CMT than I do. This is her story:

“I am a lefty. Aside from CMT, the problems with my left hand were complicated by a motor vehicle accident over ten years ago. I consulted one of my former classmates, a certified hand therapist (CHT), who works in a clinic near my home. Becoming a CHT involves specialized training and passing a national certification examination beyond the regular OT (or PT) exam.

“For three years a hand surgeon and my CHT followed me on a regular basis. Using a dynamometer we were able to objectively measure the loss of grip strength over time. Being able to demonstrate the weakness in my grasp (and sensory loss) and also how quickly my hand fatigued helped us figure out when surgery was needed. It was very startling to see when my grasp was eight pounds on the first attempt and 50% of that strength on the next two attempts. Minimum grasp for function is around twenty pounds.

“During this period three custom-made splints were constructed. The first provided external support to compensate for the weakness in my thumb and wrist. I used this splint during activities such as gardening and fishing. A second splint was constructed to provide rest and support when I overtaxed my hand. I would wear this splint during painful flares of tendonitis. The third splint (a resting hand splint which I could sleep in) kept my hand in a position that was not stressful on my joints, tendons or ligaments.

“I also used adaptive equipment and energy conservation techniques. These were very important, as my loss of strength and sensation affected what I could do during the day. The most important adaptive equipment was often for the simplest tasks—scissors that I could

manage, an adapted key to turn on my bedside lamp, built-up handles in the kitchen and garden, and adapted keys for my vehicle and front door. These all made significant improvements in the quality of my life.

“Late last year the surgeon and OT suggested tendon transfer surgery. The tendon chosen was the palmaris longus (a wrist flexor), which was “woven through” the extensor pollicis (a thumb muscle) to give me a bit of thumb extension when I extend my wrist. This helps keep the thumb out of the way. The goals of this procedure were to stabilize my wrist and decrease the pain, and hopefully gain wrist extension. Two weeks after the surgery (when I came out of my cast) I consulted my OT for a custom-made splint to immobilize the wrist while the tendons healed. During the time that I was limited to only moving my fingers, my OT worked to decrease the hypersensitivity along the surgical sites. Ultrasound and massage helped decrease the swelling. Hooking the tendon through the thumb muscle gave me a thickened area on that side of my wrist, but I am so pleased with the function that I don’t mind the bulge.

“Once the transfer tendon had healed, it was time to start strengthening the muscles and reeducating the transferred tendon. Prior to the surgery this tendon flexed my wrist. Now it had been moved to the other side and had to be trained to perform wrist extension. It took around 6 weeks before I actively tried to move the wrist. At first, I would tap on it and try to contract it. Once I was cleared for active motion, it was amazingly easy. My OT used vibration and tapping to re-train the muscles and the message went through.

“It has been five months since my surgery. Although the recovery time and the amount of pain were much more than I anticipated, I am very pleased with the results of the surgery. I have significantly less pain and stress on my wrist and thumb. A real bonus for me is that (although my hand remains very weak) I am able to extend the wrist and thumb. This enables me to use my left hand much more skillfully.”

HANDS ARE ONE FOCUS OF OCCUPATIONAL THERAPY



The Collum™ thumb support is made of leather with a metal stay.



A hand splint supplies support when the hand is at rest.



A right-angle knife requires less strength to cut.

Professionals Working for CMT Patients: Dr. James Lupski

Dr. James Lupski is the Cullen Professor of Molecular and Human Genetics and Professor of Pediatrics at Baylor College of Medicine. Dr. Lupski received his undergraduate training at New York University. During his undergraduate studies he worked as an undergraduate research participant at the Cold Spring Harbor Laboratories. The Cold Spring Harbor Laboratories is directed by Dr. James D. Watson, the co-discoverer of the “Watson-Crick Model of DNA.” It was there that Dr. Lupski learned the genetic engineering technologies that would be crucial to the identification of the first mutation and genes involved in Charcot-Marie-Tooth disease.

Dr. Lupski received his medical and graduate education at New York University School of Medicine, where he was a participant in the Medical Scientist Training Program. His Ph.D. was in bacterial genetics.

Dr. Lupski is one of eight children, four of whom are affected with Charcot-Marie-Tooth disease. His parents are unaffected, which suggests recessive inheritance. During his teenage years, he underwent a number of orthopedic procedures for correction of severe pes cavus and other foot deformities. It is during this time that his interest in Charcot-Marie-Tooth disease research and genetics was greatly stimulated.

After completing the M.D./Ph.D. Program at New York University, Dr. Lupski was named to the faculty of Biochemistry as a Research Assistant Professor. It was during this time that the famous polymerase chain reaction (“PCR”) was discovered. It became clear that genetic engineering experiments, which were done only in bacteria up to this time, would be readily applicable to the study of human medical genetic conditions. Dr. Lupski then did his residency training in pediatrics as well as a medical genetics fellowship at Baylor College of Medicine. This institution was chosen because it ranks among the top three medical genetics institutions in the United States, with one of the top pediatrics training programs.



After the discovery in 1984 of a method to map genes responsible for human genetic disease and, in particular, Huntington disease, Dr. Lupski planned to utilize these approaches to identify the first Charcot-Marie-Tooth disease genes upon completion of his training at Baylor College of Medicine. It was during his residency and genetics fellowship training that Dr. Lupski was also appointed to the faculty in the Department of Genetics at Baylor College of Medicine. He completed his fellowship in 1991,

which was the same year that, in collaboration with Dr. Pragna Patel, Dr. Lupski's group described the Charcot-Marie-Tooth disease type 1A duplication as a novel molecular mechanism for human genetic disease. In 1995, Dr. Lupski was promoted to Full Professor and awarded the Cullen endowed professorship in the Department of Molecular and Human Genetics. His laboratory has worked extensively on the genetics of Charcot-Marie-Tooth disease and has been responsible

for identifying a number of the genes and mutational mechanisms, developing diagnostics for clinical application, and collaborating with patients and families who have CMT disease or a related peripheral neuropathy using molecular approaches to understand the disease process. A major part of the collaborations has been done in conjunction with Dr. Carlos A. Garcia, working with families from the MDA Clinics in New Orleans, Baton Rouge, and Lafayette, Louisiana. To date, the Lupski laboratory is responsible for close to 100 publications on the molecular genetics and clinical studies of Charcot-Marie-Tooth disease and related neuropathies.

Dr. Lupski states that “having Charcot-Marie-Tooth disease has certainly been a major force in the research efforts to try to understand the molecular genetic bases of the disease, develop diagnostics to molecularly define the disorder, and hopefully, through a better understanding, to develop treatments which will be directed at the underlying cause and not just symptomatic relief.”

Dr. Lupski has been a member of the CMTA Medical Advisory Board since 1990.

“The CMTA and patient support groups like it provide patients with useful clinical information, advocate for patients and support the research which will lead to a better understanding and better treatments of CMT.”

—Dr. James Lupski



VISIT OUR HOMEPAGE

at

www.charcot-marie-tooth.org

The Genetic Testing Lawsuit from the

By **MAUREEN P. HORTON, R.N.**

(This is the second installment of an article concerning genetic testing in the workplace. Here, Maureen Horton considers the implications of the decisions in the Burlington Northern Santa Fe Railway [BNSF] case on people with genetic diseases.)

One would be hard pressed to find any physician—outside of Burlington Northern, that is—who thinks that it is okay to test a person's genes without his or her knowledge or consent. It just is not done. It is not ethical. But beyond informed consent, what are the issues for us?

To be politically correct, or maybe just kind, let's say that this testing shows that a little knowledge can be a dangerous thing—and in the case of BNSF, expensive, too. There is a disease that causes carpal tunnel syndrome (CTS), in the majority of the patients who are known to have the disease. And there is a genetic test for the disease. The disease is called HNPP. But one only needs a very basic understanding of HNPP, to realize how absurd testing was.

First the numbers: CTS is an extremely common repetitive motion injury. Everyone knows at least someone with CTS. HNPP is rare. HNPP is estimated to affect 1:2,500 to 1:6,000 people. In a city with a population of 40,000 (the size of the BNSF workforce) one would statistically expect to find 7 to 16 people. Many researchers think that the majority, some say as high as 90% of those with HNPP, are not yet diagnosed. So in this fictitious town, maybe only 1 to 3 people are diagnosed with HNPP. And since it is an inherited disorder, they are likely to be related. In testing all its employees who were filing claims and were diagnosed with CTS, BNSF was searching for the proverbial needle in the haystack.

HNPP is marked by a combination of symptoms and abnormal test results, of which CTS is only one problem. Dr. Michael Shy stated, "I personally know of no HNPP patient whose only abnormality on nerve conduction testing is a CTS. While there is variability in nerve conduction velocity with HNPP, I have never seen or heard of CTS being an isolated finding. Thus the premise of the company was incorrect in this context as well as being improper." Testing for HNPP in a carpal tunnel patient is appropriate if there is a family history suggesting HNPP, presence of a generalized demyelinating neuropathy

or signs on physical exam suggesting HNPP (i.e. pes cavus, depressed deep tendon reflexes, etc.)," according to Dr. Phillip Chance.

Even if BNSF had found someone with a positive DNA test, it would tell little more than the fact that the person had HNPP. HNPP symptoms are highly variable. A positive test is no indication of when a person will become symptomatic, how 'bad' the symptoms will be, how often they will recur, nor which parts of the body will be involved. Given the intermittent nature of the pressure palsies, even a bad episode or two is no predictor of future events. Individuals can go years, even decades, between episodes while pursuing careers or hobbies involving strenuous activity. The only predictor we have right now is time. Seeing a pattern of frequent or severe pressure palsy episodes—or lack of them—over decades will give the doctors some loose indication of what is to come. One test or one episode says little.

We know that pressure palsies are—in part—caused by repetitive activities. With some companies looking at work site ergonomics and looking for ways to lower the incidence of repetitive motion injuries, is it ever appropriate for them to do such testing? Dr. Thomas Bird says no. "Companies shouldn't be in the business of ordering genetic tests on their employees with various symptoms or illnesses. This should be done by physicians with appropriate expertise. Of course, companies can hire physicians and tell them what to order, but that's another problem." Dr. Bird went on to say, "There seems to be no reason to order the HNPP test on just anyone with CTS. The test would be more appropriate if the patient had recurrent CTS, or multiple unexplained compression neuropathies, or especially if there were a documented positive family history of other persons with CTS. Even then, we have seen persons with familial CTS turn out to have CMT (such as CMT1B) rather than HNPP. And, of course, there can be recurrent CTS and familial CTS and no genetic evidence for HNPP or CMT. So it gets pretty complicated."

In the HNPP community, it is not at all uncommon to have family members showing signs of HNPP but refusing to be tested. Some don't want to think about it or what it may mean. For now, denial works for them. Others, very intentionally, want to delay a diagnosis as long as possible. They are concerned, and rightly so, about their careers and insurance. The ADA,

HNPP/CMT Community Perspective...

the Civil Rights Act, the Age Discrimination in Employment Act, and the Equal Pay Act* have all been enacted to guard against discrimination. But in reality, we know that discrimination happens every day.

Having the diagnosis and trying to get a first job, maintain a job, or change employment is of great concern to many. A human resources person, who asked to remain anonymous, says there is no way to know for sure if disclosing a potentially disabling condition up front (on application or during the interview) will help or hinder the chances of being hired. If the person doing the interviewing and hiring has a minority quota to fill, or has a family member with a disability or has had a positive experience with a disabled employee, then discrimination is less likely. In this case it is favorable to disclose the condition. But the opposite is also true. It is difficult to prove that you were not hired because you disclosed a potentially disabling condition. Once you are hired, reasonable accommodations have to be made. But "reasonable" is arbitrary. Some companies will do much more to help an employee stay in the job than others.

People are also concerned, and rightly so, about insurance. Health insurance, long-term care and long- and short-term disability insurance can exclude existing conditions entirely or for a specific time period or pay out at a lesser rate. This is one time to read the fine print whether you are diagnosed or not. Dr. Shy wrote, "I share the concern of family members who think they probably have HNPP but don't want testing for insurance reasons, etc. Our feeling is that once a patient in the family has positive genetic testing results, it is usually not necessary to perform genetic testing on other family members to see if they are affected. This can usually be done by a competent physician's exam and NCV testing; then there are no genetic testing results on their chart. This is an important ethical issue. This is one reason why we take extreme steps to ensure that patient confidentiality is respected and identifiers are removed for our database and why we send results of our clinical analysis to the patients themselves so that they can distribute our conclusions to whom they feel it will be most appropriate."

So how do we take the good of knowing our gene make-up and avoid the exploitation of that knowledge? Dr. Bird says, "In terms of legislation, a good start is to prevent insurance com-

panies from using genetic test results to deny insurance. You know we all have genetic mutations, and eventually that will be shown by future tests, and we will all be more or less in the same boat. Also companies (employers) should not discriminate on the basis of genetic tests, unless there is clear documentation of a potential harm to the employee in that particular occupation. Presumably someone at an increased genetic risk for cancer should not be working around cancer-causing chemicals (but then who should be?)."

In the case of BNSF, several of its employees have begun work alongside Senator Tom Daschle and Congresswoman Louise Slaughter on Capital Hill to urge legislation banning genetic discrimination in the workplace and from the health insurance industry. President Bush announced his support for the idea. Many expect Bush will push for a weaker version of such a law after lobbying by the employer and insurance industry groups. With a democratic majority in the Senate, action on the Daschle-Slaughter bill is expected this year.

Quoted in the Amarillo Globe article, Dr. Jeffrey P. Kahn, director of the Center for Bioethics at the University of Minnesota, said the BNSF testing program was "sort of a wake-up call" for the public about genetic testing. The tests for CMT and HNPP exist. This is not a 'sort of,' but an honest-to-goodness wake-up call. Right now. We are at the forefront of genetic testing ethics. This issue involves us, our children and grandchildren. Now is the time to get involved. Call, or preferably, write your local and state senators and representatives. Tell them your own story. Share what has happened to you. Emphasize the need to ban genetic testing discrimination in the workplace and insurance industry. Do it now.

** In addition, EEOC enforces Title VII of the Civil Rights Act of 1964, which prohibits discrimination on the bases of race, color, religion, sex or national origin; the Age Discrimination in Employment Act, which protects workers age 40 and older; and the Equal Pay Act, which prohibits sex-based differences in compensation. Further information about EEOC, including its ADA policy guidances, is available on the agency's Web site at <http://www.eeoc.gov/>.*

Note: Statements from doctors via e-mail

HNPP website: <http://www.hnpp.org>

New Lightweight Brace Makes Walking Easier

By **WILLIAM B. QUINN, DPM, FACFAS**

The Walk Right brace is a new innovative, comfortable, drop-foot assistance device that will help make walking easier. This new walking assistance can be attached to any style tie shoe. Keith Markusic, the president of Walk Right, originally designed the brace for his own use, as he has a drop-foot problem himself. Frustrated with the problems of conventional AFOs (ankle-foot orthotics), Keith developed a new brace that is lightweight, non-rigid and comfortable. The brace is attached to the outside of any tie shoe (including athletic shoes), allowing the individual to wear his or her actual shoe size and not oversized shoes to accommodate a brace. This also eliminates brace irritation and allows the cushioned insert of the shoe to comfort the foot. Once the brace is fitted and comfortable, all other shoes can be modified to accept the brace, allowing multiple shoe styles to be worn.



Because of the different levels of disability that CMT can cause, this brace may not help individuals with very strict, rigid support requirements. However, the brace is ideal for mild to moderate foot drop and for those individuals requiring minimal bracing. The brace is dynamic in function and allows for continuing muscle function, thus reducing muscle atrophy that usually accompanies bracing.

Compared to standard AFO bracing, the cost of the Walk Right brace is reasonable. The cost of the brace is \$150 per brace, or \$300 for a pair, which is less than half the cost of conventional AFOs. Convenience is also an issue with getting braces made. The Walk Right brace is measured at home and sold through the mail, thus eliminating the inconvenience of multiple clinic visits and helping to reduce the cost.

ADVANTAGES OF WALK RIGHT BRACE

- Lightest of all AFOs and foot-drop braces.
- Brace can be attached to any tie shoe.
- Affordably priced.
- The brace is cosmetically fashioned and unrecognizable under long pants.
- No plastic or other hard insert is placed on the inside of your shoes.
- It is no longer necessary to buy two different size shoes to wear your brace.
- Highly recommended for diabetic patients with neuropathy and those patients with foot sensory alteration, because the brace does not require any hard insert into the shoe.
- Prevents foot ulceration, while providing a cushioned area (the natural inside of the shoe) for protection and healing of existing foot ulcers.
- Lightweight and cool for hot weather.
 - Provides for a more even and normal gait when walking. The elastic bands provide passive resistance against the foot, allowing a more normal gait.
 - Increases the level of physical activity in most patients.
 - Many individuals can return to sports and activities they had to set aside because of their foot drop.
- Walking up hills and inclines is much easier because of the patented design.
- The brace is easier for the patient or caregiver to put on than the current AFOs.
- The patient in the hospital setting can now benefit from the brace. The physical therapist can start patients ambulating while in house by having a selection of shoes with the brace adapters on them and a selection of braces for them to wear.
- The brace can prevent foot drop on patients on prolonged bed rest and inactivity by applying the shoes and braces while they are in bed. The patented design makes the brace comfortable while the wearer is lying in bed and provides enough tension that the patient can utilize the resistance of the elastic bands to perform flexion and extension of the foot.

Keith Markusic, who is the president of Walk Right, Inc., uses the Walk Right brace every day. If you have any questions, please feel free to call him at any time. He will follow up all inquiries quickly. You may call him directly. His personal home/office phone number is 330-757-8613.

The Year in Review:

Fiscal Year, July 1, 2000 to June 30, 2001

By **ARDITH FETTEROLF, President**

The CMTA is moving forward at a fast pace. Our financial health is the best it has ever been, thanks in part to our membership giving, donor pledges, bequests, support for the new North American Database, and our special fundraising events, such as our golf tournaments.

Our new office facility provides an improved physical environment, and is well-suited to the outstanding performance of our small staff as they meet the increasing demands placed upon them.

Our Board members and our Director of Program Services were instrumental in organizing and attending a number of major CMTA functions as well as medical conventions. In the past year, we have sponsored three patient family conferences at the University of Utah, the University of Pennsylvania and the most recent at Rancho Los Amigos in Downey, California. We had planned one for Tulane University in New Orleans which was, unfortunately, cancelled due to low registration.

We successfully planned and subsidized a national gathering of CMTA support group leaders in St. Louis, Missouri, which was enthusiastically attended by the majority of our group leaders. The purpose of the meeting was to galvanize the work done at the local level and to make each group feel more like an arm of the national organization. That mission was accomplished!

Chairman Ann Lee Beyer staffed booths at a number of medical conventions, including the American Neurological Association, the American Academy of Neurology and the Human Genetics Society. I attended the American Podiatrists Association meeting so more shoe fitters would be familiar with our organization. Both Ann and I attended a three-day conference of the Mediterranean Basin

Neuromuscular Society on the Isle of Capri, which was funded by private donors. Many of the Board of Directors attended the first Young Investigators Meeting, which took place in Boston during the ANA meeting, where grant recipients presented the findings of their CMTA research work.

One of the major accomplishments of this fiscal year was the awarding of five post-doctoral grants by the association. The topics were diverse and so were the recipients, ranging from genetic studies at Baylor to practical therapy treatments by a group in Australia. A second major accomplishment in CMT research was the formation of the North American Database at Indiana University to which the CMTA has made a three-year financial commitment. Information about CMT patients and families will be stored there for use by researchers around the world. The hope is that this storehouse of information will speed up the work of researchers looking at both the clinical and genetic aspects of CMT.

In an effort to improve the way the association works and serves its members, three Board members attended the National Organization of Non-Profit Boards seminar in Washington, DC. Additionally, the Board meets once a quarter to manage the work of the organization and to plan its goals for the upcoming months.

The biggest undertaking of recent months has been a new method of billing for membership. Previously, we billed everyone in the summer months and it was confusing to people who had joined at other times of the year. Now, we will bill each person in the month he joins or renews his membership. Initially, it has created a lot of work for the office, but eventually, it should make the whole process much more efficient.

We thank all of you, our members, for your ongoing support of our efforts on your behalf. Without you, there would be no CMTA.

REHABILITATION MANAGEMENT OF CHARCOT-MARIE-TOOTH DISEASE PUBLISHED

Paolo Vinci, M.D. has recently published a small volume on rehabilitation options for patients with CMTA and rehabilitation professionals. Dr. Vinci does, himself, have CMT and decided to write a text based on both his experiences as a patient and as a physiatrist, specializing in neuromuscular rehabilitation.

The text is divided into three parts: Primary Anatomophysiology of the Motor System, Pathogenesis of Charcot-Marie-Tooth Disease, and the largest section, Rehabilitation Management. The text is small (about 120 pages), but the book is filled with full-color photos and very detailed diagrams on such things as gait, normal stance, and finger grip.

The book was published in Italy and can be ordered by credit card from the website at www.stradeservizi.it/cmt.htm. The cost is approximately \$38.00 in US funds, plus postage and handling.

Charcot-Marie-Tooth Association

INDEPENDENT AUDITORS' REPORT

To the Board of Directors
Charcot-Marie-Tooth Association
Chester, Pennsylvania

We have audited the accompanying statement of financial position of Charcot-Marie-Tooth Association (a Pennsylvania nonprofit corporation) as of June 30, 2001, the related statements of activities, cash flows and functional expenses for the year then ended. These financial statements are the responsibility of the Association's management. Our responsibility is to express an opinion on these financial statements based on our audit. The prior-year summarized comparative information has been derived from the Organization's 2000 financial statements and, in our report dated August 11, 2000, we expressed an unqualified opinion on those financial statements.

We conducted our audit in accordance with auditing standards generally accepted in the United States. Those standards require that we

plan and perform the audit to obtain reasonable assurance about whether the financial statements are free of material misstatement. An audit includes examining, on a test basis, evidence supporting the amounts and disclosures in the financial statements. An audit also includes assessing the accounting principles used and significant estimates made by management, as well as evaluating the overall financial statement presentation. We believe that our audit provides a reasonable basis for our opinion.

In our opinion, the financial statements referred to above present fairly, in all material respects, the financial position of Charcot-Marie-Tooth Association at June 30, 2001, and the changes in its net assets and its cash flows for the year then ended, in conformity with generally accepted accounting principles.



August 13, 2001

STATEMENT OF FINANCIAL POSITION

JUNE 30, 2001 WITH COMPARATIVE TOTALS FOR JUNE 30, 2000

| ASSETS | 2001 | 2000 |
|--|------------------|------------------|
| CURRENT ASSETS | | |
| Cash | \$457,932 | \$343,403 |
| Unconditional promises to give | 55,000 | - |
| | <u>512,932</u> | <u>343,403</u> |
| EQUIPMENT - Net of accumulated depreciation of \$11,719 | 15,616 | 17,746 |
| OTHER ASSETS | 4,733 | 4,523 |
| TOTAL ASSETS | <u>\$533,281</u> | <u>\$365,672</u> |
| LIABILITIES AND NET ASSETS | | |
| CURRENT LIABILITIES | | |
| Accounts payable and accrued expenses | \$ 6,584 | \$ 2,612 |
| NET ASSETS | | |
| UNRESTRICTED | 344,539 | 164,206 |
| TEMPORARILY RESTRICTED | 182,158 | 198,854 |
| TOTAL NET ASSETS | <u>526,697</u> | <u>363,060</u> |
| TOTAL LIABILITIES AND NET ASSETS | <u>\$533,281</u> | <u>\$365,672</u> |

STATEMENTS OF CASH FLOWS

JUNE 30, 2001 WITH COMPARATIVE TOTALS FOR JUNE 30, 2000

| | 2001 | 2000 |
|--|----------------|----------------|
| CASH FLOWS FROM OPERATING ACTIVITIES | | |
| Change in net assets | \$ 163,637 | \$ 163,881 |
| Adjustments to reconcile change in net assets to net cash provided by operating activities | | |
| Depreciation | 5,122 | 4,613 |
| Donated equipment | (700) | (1,660) |
| (Increase) decrease in assets | | |
| Unconditional promises to give | (55,000) | 54,250 |
| Other assets | (210) | (2,635) |
| Increase (decrease) in liabilities | | |
| Accounts payable and accrued expenses | 3,972 | (3,694) |
| Net cash provided by operating activities | <u>116,821</u> | <u>214,755</u> |
| CASH FLOWS FROM INVESTING ACTIVITIES | | |
| Purchase of property and equipment | (2,292) | (3,506) |
| NET INCREASE IN CASH | 114,529 | 211,249 |
| CASH - BEGINNING OF YEAR | 343,403 | 132,154 |
| CASH - END OF YEAR | <u>457,932</u> | <u>343,403</u> |

The accompanying notes are an integral part of these financial statements. See page 14.

Financial Report as of June 30, 2001

STATEMENT OF ACTIVITIES

YEAR ENDED JUNE 30, 2001

(WITH SUMMARIZED FINANCIAL INFORMATION FOR THE YEAR ENDED JUNE 30, 2000)

| | 2001 | | | 2000 |
|--|------------------|---------------------------|------------------|------------------|
| | UNRESTRICTED | TEMPORARILY RESTRICTED | TOTAL | TOTAL |
| SUPPORT AND REVENUES | | | | |
| Contributions | \$345,225 | \$211,853 | \$557,078 | \$442,873 |
| Special fundraising events | 72,795 | - | 72,795 | 18,050 |
| Cost of special events | (30,939) | - | (30,939) | (2,915) |
| Interest income | 23,979 | - | 23,979 | 9,550 |
| Donated equipment | 700 | - | 700 | 1,660 |
| | <u>411,760</u> | <u>211,853</u> | <u>623,613</u> | <u>469,218</u> |
| NET ASSETS RELEASED FROM RESTRICTIONS | | | | |
| Satisfaction of program restrictions | 228,549 | (228,549) | - | - |
| TOTAL SUPPORT AND REVENUES | <u>640,309</u> | <u>(16,696)</u> | <u>623,613</u> | <u>469,218</u> |
| EXPENSES | | | | |
| Program services | 386,898 | - | 386,898 | 253,992 |
| Fundraising | 32,426 | - | 32,426 | 15,775 |
| Management and general | 40,652 | - | 40,652 | 35,570 |
| TOTAL EXPENSES | <u>459,976</u> | <u>-</u> | <u>459,976</u> | <u>305,337</u> |
| CHANGE IN NET ASSETS | 180,333 | (16,696) | 163,637 | 163,881 |
| NET ASSETS - BEGINNING OF YEAR | <u>164,206</u> | <u>198,854</u> | <u>363,060</u> | <u>199,179</u> |
| NET ASSETS - END OF YEAR | <u>\$344,539</u> | <u>\$182,158</u> | <u>\$526,697</u> | <u>\$363,060</u> |

The accompanying notes are an integral part of these financial statements. See page 14.

STATEMENT OF FUNCTIONAL EXPENSES

YEAR ENDED JUNE 30, 2001

(WITH SUMMARIZED FINANCIAL INFORMATION FOR THE YEAR ENDED JUNE 30, 2000)

| | 2001 | | | 2000 |
|----------------------------------|---------------------|------------------|---------------------------|-------------------|
| | PROGRAM SERVICES | FUNDRAISING | MANAGEMENT AND GENERAL | TOTAL EXPENSES |
| Salaries and benefits | \$ 38,207 | \$ 2,090 | \$ 24,512 | \$ 64,809 |
| Publications and supplies | 17,654 | 30,336 | - | 47,990 |
| Occupancy and office expense | 53,570 | - | 10,204 | 63,774 |
| Research fellowships | 232,181 | - | - | 232,181 |
| Conference expense | 39,327 | - | - | 39,327 |
| Consulting | 1,924 | - | - | 1,924 |
| Professional fees | 194 | - | 4,655 | 4,849 |
| Depreciation | 3,841 | - | 1,281 | 5,122 |
| TOTAL FUNCTIONAL EXPENSES | <u>\$386,898</u> | <u>\$ 32,426</u> | <u>\$ 40,652</u> | <u>\$459,976</u> |
| | | | | <u>\$305,337</u> |

The accompanying notes are an integral part of these financial statements. See page 14.

CHARCOT-MARIE-TOOTH ASSOCIATION NOTES TO FINANCIAL STATEMENTS—JUNE 30, 2001

NOTE 1 - SUMMARY OF SIGNIFICANT ACCOUNTING POLICIES

Nature of Operations

The Charcot-Marie-Tooth Association (the "Association") was established to create awareness of Charcot-Marie-Tooth ("CMT") disorders within the health care community and the general public, and be a leading source of information regarding CMT disorders. The Association encourages, promotes and supports research into the cause, treatment and cure of CMT. The Association also facilitates education and support for persons affected by CMT.

Basis of Presentation

The Association follows Statement of Financial Accounting Standards (SFAS) No. 117, Financial Statements of Not-for-Profit Organizations to prepare its financial statements. Under SFAS No. 117, the Association is required to report information regarding its financial position and activities according to three classes of net assets: unrestricted net assets, temporarily restricted net assets and permanently restricted net assets.

Restricted and Unrestricted Support

The Association follows SFAS No. 116, Accounting for Contributions Received and Contributions Made in recording contributions received. Contributions received are recorded as unrestricted, temporarily restricted, or permanently restricted support, depending on the existence and/or nature of any donor restrictions.

Support that is restricted by the donor is reported as an increase in unrestricted net assets if the restriction expires in the reporting period in which the support is recognized. All other donor-restricted support is reported as an increase in temporarily or permanently restricted net assets, depending on the nature of the restriction. When a restriction expires (that is, when a stipulated time restriction ends or purpose restriction is accomplished), temporarily restricted net assets are reclassified to unrestricted net assets and reported in the Statement of Activities as net assets released from restrictions. There are no permanently restricted funds.

Estimates

The preparation of financial statements in conformity with generally accepted accounting principles requires the use of estimates based on management's knowledge and experience. Accordingly, actual results could differ from those estimates.

Functional Allocation of Expenses

The costs of providing the various programs and other activities have been summarized on a functional basis in the statement of activities. Accordingly, certain costs have been allocated among the programs and supporting services benefited.

Equipment and Depreciation

Equipment is recorded at cost. Depreciation is provided on a straight-line basis over the estimated useful lives of the assets.

Tax Status

The Association is incorporated in the Commonwealth of Pennsylvania and is exempt from federal income taxes

under Section 501(c)(3) of the Internal Revenue Code. The Association is registered as required with the Pennsylvania Bureau of Charitable Organizations.

Unconditional Promises to Give

Unconditional promises to give represents payments due in future periods for awards recorded as temporarily restricted support and revenue.

In-kind Contributions

Volunteers have donated their time to the Association's program and administrative services and its fund raising campaigns. The value of this contributed time is not reflected in these statements since it is not susceptible to objective measurement or valuation.

The Association occasionally receives donations of property and equipment. During the year ended June 30, 2001, the value of donated assets is \$700 and is reflected in the Association's funds.

Prior Year Comparative Data

The financial statements include certain prior-year summarized comparative information in total but not by net asset class. Such information does not include sufficient detail to constitute a presentation in conformity with generally accepted accounting principles. Accordingly, such information should be read in conjunction with the Association's financial statements for the year ended June 30, 2000, from which the summarized information was derived.

NOTE 2 - CONCENTRATION OF CREDIT RISK

During the year the Association may have deposits with major financial institutions which exceed Federal Depository Insurance limits. These financial institutions have strong credit ratings, and management believes the credit risk related to these deposits is minimal.

NOTE 3 - TEMPORARILY RESTRICTED FUND

At June 30, 2001, the temporarily restricted fund had a balance of \$182,158 comprised of monies for research grants and education.

NOTE 4 - LEASES

The organization conducts its operations from a facility leased under an operating lease expiring in November of 2002. At June 30, 2001, the organization was obligated under this lease arrangement as follows:

| YEARS ENDING | AMOUNT |
|--------------|-----------------|
| JUNE 30, | |
| 2002 | 17,500 |
| 2003 | 7,500 |
| | <u>\$25,300</u> |

NOTE 5 - TAX-DEFERRED ANNUITY PLAN

The Association has a tax-deferred annuity plan qualified under Section 403(b) of the Internal Revenue Code. The plan covers full-time employees of the Association. The Association contributes 3% of gross salaries for qualified employees to the plan. Plan expenses were \$824 for the year ended June 30, 2001.

The Herb of the Month: St. John's Wort

By **BRUCE A. CRISTOL, Pharm.D.**

At a recent family conference of the Charcot-Marie-Tooth Association at Rancho Los Amigos National Rehabilitation Center in Downey, California, I had the privilege of presenting a modest contribution on the effects of medications when given together with other medications, commonly known as drug-drug interactions. We in the Pharmacy Department at Rancho encounter these concerns regularly. Following the presentation a question and answer session occurred, and the issue of the impact of herbs on the medications taken by CMT patients was addressed by an individual in the audience. As a result I am hoping that CMTA members may benefit by becoming more aware of how specific herbs can influence the effectiveness of their medications; in other words, it is important to be aware that herbs may cause problems when given in combination with certain medications.

St. John's wort is a fragrant plant (weed) found in the dry ground of roadsides, meadows, woods, and hedges. Usually growing to a height of one to two feet in the United States and Canada, on the Pacific Coast it has been found at heights of up to five feet. With oval-shaped leaves and golden-yellow flowers (blooming

from June to September), the plant is harvested for medicinal purposes in the months of July and August. In addition, to retain potency it must be dried immediately.

St. John's wort has been used as an herbal remedy since the Middle Ages for a number of ailments, but fell into disuse until recent times. In the past fifteen years it has become a component of many herbal preparations for the treatment of anxiety and depression. Medical studies have shown that the active ingredients in St. John's wort are effective in mildly to moderately depressed individuals.

Of concern to both patients and pharmacists are the following: (1) St. John's wort is not considered a "drug" by the U.S. Food and Drug Administration (FDA). Therefore, because standards have not been set, the concentration of the active ingredients can vary from dose to dose, batch to batch, or manufacturer to manufacturer; (2) there are drug-herb interactions which have been uncovered in the past several years which the patient should be aware of.

Until St. John's wort is classified as a drug by the FDA, standardized dosage forms cannot be assured. However, the patient can become aware of the drug-herb interactions reported in the medical literature. They are classified as shown in the chart below.

| Drug Interactions with St. John's Wort | Result of Interaction | Comments |
|--|---|--|
| Paroxetine (Paxil™) | Lethargy/incoherence | |
| Trazodone (Desyre™) Sertraline (Zoloff™) Nefazodone (Serzone™) | Serotonin syndrome* Serotonin syndrome* Serotonin syndrome* | A similar case has been documented with St. John's wort alone. |
| Theophylline Digoxin (Lanoxin™) Cyclosporin (Sandimmune™) | Decreased theophylline concentrations Decreased digoxin concentrations Decreased cyclosporin concentrations | St. John's wort has been shown to affect the concentrations in the blood of these medications because it prevents the liver (which helps rid the body of the medications) from functioning as efficiently as it would otherwise. |
| Oral contraceptives | Breakthrough bleeding | |
| Iron products | Impaired absorption | Impaired iron absorption is more of a theoretical concern. It may or may not occur. |

* symptoms include: changes in mental status; shaking; stomach upset; headache; muscle pain; restlessness

REFERENCES:

- DerMarderosian, A. The Review of Natural Products. *Facts and Comparisons* 1999.
 Ernst, E. Second thoughts about safety of St. John's wort. *Lancet* 1999; 354:2014-2016.
 Fugh-Berman, A. Herb-drug interactions. *Lancet* 2000; 355:134-138.
 Miller, L. Herbal medicinals: selected clinical considerations focusing on known or potential drug-herb interactions. *Archives of Internal Medicine* 1998; 158:2200-2211.

UPCOMING CONFERENCE

The next Patient/Family Conference is scheduled for November 10, 2001 at the University of Miami. Information will be mailed to all Florida residents. Members outside of Florida interested in attending can call the office at 1-800-606-CMTA for a registration form.



Support Group News

■ California—Berkeley Area

The support group held its most recent meeting on September 15, 2001, from 2-4 PM in the Albany, CA, library. The group is promoting the CMT North American Database and encouraging members to fill out the questionnaires. Additionally, the group has decided to meet quarterly during 2002, with planned meetings in January, April, July and October.

■ California—North Coast Counties

The next meeting of this group will be November 4, 2001. They, too, are encouraging their members to complete the database survey forms and return them to help supply information for the researchers. Their newsletter featured the following ideas about exercise from a presentation by Dr. Joan Dahmer:

1. People with peripheral neuropathy benefit from aerobic exercise as much as everyone else.
2. If the nerve supply to a muscle is compromised by CMT, this cannot be overcome by aerobic exercise.
3. You can improve muscle bulk in non-affected or even less affected muscle fiber with exercise.
4. If you can't feel your feet, you must be very careful about injuring them in recurrent weight bearing.
5. The more you use your body's systems for balance, by walking, the better shape you will be in.

Greenville Ohio Support Group members (left to right): Joe Whitacre, Dot Cain, Gayle Emery, Peg Willis and Delores Houston.



The Benson, Minnesota support group gathered for a picture following their last meeting.

6. Weight bearing is good for bones. The weaker your bones, the more likely they are to break if you fall, and CMT increases your risk of falling.
7. Exercise is a natural way to combat stress.

■ Kentucky/Southern Indiana/Southern Ohio

The last meeting was held on September 15, 2001 at the Northside Branch of the Lexington Public Library. The program centered around two presentations: Independence Living Centers, whose mission is to educate and inform people about the need of the disabled to be as responsible as possible for their own needs, and Benefits Plus, a state program of Social Security, which helps beneficiaries understand their work options while on SSI or SSDI.

■ Minnesota—Benson

The last meeting of the support group discussed the national support group leaders' conference and the speech given there by Dr. Florian Thomas. Eleven people attended who have CMT with five spouses. One man traveled four hours to attend the meeting. The group gathered for a photo following the meeting.

■ Ohio—Greenville

At the last meeting of the CMT support group at the Church of the Brethren in Greenville, OH, the attendees posed for a picture showing the many forms of AFOs they use to increase their mobility.

CMTA Support Groups

Arkansas—Northwest Area

Place: Varies, Call for locations
Meeting: Quarterly
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: CMT_losangeles@yahoo.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

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—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: Saint Louis University Hospital
Meeting: Quarterly
Contact: Carole Haislip, 314-644-1664
E-mail: c.haislip@att.net

New York—Greater New York

Place: NYU Medical Center/
Rusk Institute, 400 E. 34th St.
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: Greenville-Ohio-CMT@
woh.rr.com

Oregon/Pacific NW

Place: Portland, Legacy Good Sam
Hospital, odd months
Brooks, Assembly of God
Church, even months
Meeting: 3rd Saturday of the month
(except June and Dec.)
Contact: Jeanie Porter, 503-591-9412
Darlene Weston, 503-245-8444
E-mail: jeanie421@yahoo.com or
blzerbabe@aol.com

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

**GENEROUS
BEQUEST
RECEIVED**

A \$40,000 bequest was received from the estate of Dorothy Anne Ferenbaugh.

GIFTS WERE MADE TO THE CMTA

IN MEMORY OF:

Marguerite Baker
Mr. & Mrs. Dennis Stanley

Marjorie Bank
Carol Alsup & Pauline Beetem
Katherine Blevins
Geraldine Byrne
Hada Woodard Haynie
Jane Joyce
Mr. & Mrs. Fred Linneman
Nellie McCaw
Lois Mitchell
Louise Ness
Randall Piserchio
Leonard & Linda Walmsley
David Woodard
Helen Woodard
Lee R. Woodard
Irene & Janeen Wright
Mary & Erwin Wybenga

Mickey Donavan
Deborah Militello

Mary Guillot
Marva Barnett
Jamie & Margie Breeden
Mr. & Mrs. Peter Roane
Jack & Madolyn Steyer

John Hill
Pauline Barnes
Brian & Bona Beckley
John Dye
Eileen Goodfellow
Laura Higgins
Frederick & Constance Hubbell
Anabelle Smith
Sons of the American Legion

Grace Klein
Philip & Dorothy Dowden
Mr. & Mrs. James Findley
Elizabeth Halpern
Bill & Betsy Holloway
Ruth Ingold
Clyde & Carolyn Smith

Irvin W. Miller
Al & Connie Anderson
Frances Anderson
Mr. & Mrs. Richard Arvidson
Irene Baardsgaard
Mr. & Mrs. Dick Bautch
Jon & Bonnie Bergquist
Bill & Alice Bishop
Burdette & Doris Boyum
Dean & Darlene Conard
Ron & Laurie DeLaitre
Harold & Verneil Eickholt
Mr. & Mrs. Dan Elmer
Duane & Candis Fancher
Jeff & Nancee Frybarger
Sherry Goldenberg
Alice Gronning
Mr. & Mrs. Lou Hanus
Mr. & Mrs. Greg Jones
Mr. & Mrs. Joe Jones
Mike & Elaine Kleinprintz
Bob & Mary Kurtz
Employees of LAI Midwest
Mrs. Helen Larson
Mr. & Mrs. Gene Lillemon
Pat & Pati Moylan
Marlene Nelson
Russ Nelson
Al & Barb Peterson
Bob & Valerie Ritchie
Marlin & Rita Roehrl
Jim & Carol Rubino

Mr. & Mrs. Jack & Leona
Schneider

Don & Barb Snyder
Lorraine Stinley
Greg & Sandy Stofer
Mr. & Mrs. Arthur Strike
Ms. Marlys Sutherland
Lee & Joanne Titus
Bill & Sharon VanHorrick
Jack & Leona Vaughn
Louise Wahlberg
Connie Walbrun
Evie & Ralph Wolf
Brad & Nancy Wucherpennig
David & Julie Young
Adeline Zappa
Bob & Lynn Zauner
Barbara Zeltinger

Bessie Moring
Centerville Methodist Church

Walter Moser
Myron & Dianne Martenson
Dan & Lydia Moser

Rebecca Sand
Rhoda & Stephen Sand

Elaine Sherman
Doris & Jean Banchet
Sam Dolnick

Marjorie Spragg
Bill & Anita Harbour
Bob & Daria Homchick
Ted & Phyllis Homchick

IN HONOR OF:

Cathy Brooks
Mr. & Mrs. Kenneth Dunham

**Irene & Gerard Cavanaugh's
50th Anniversary**
Chris & Amy Burford
Frederick & Cheryl Burford
Gilbert & Ardith Burford
William & Katherine Burford
Michael & Deborah Mazzoni
Mary Beth Tech

Kent Dagg
Herbert & Frances Dorfman

Stephanie DiCara
Lillian & Ludvig Coco

Alan & Shirley Garmer
Ranee & Al Locke

Mr. & Mrs. James Goldsmith
Minna & Hart Wurzburg

Jim Howard
Sandra Howard

Charles R. Reeves
Doug Reeves

Catherine Salerno
Dr. & Mrs. James Trezza

Michael Traczuk
Ryan & Cara McElderry

CMTA Remembrances

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

Honorary Gift:

In honor of (person you wish to honor)

Send acknowledgment to:

Name: _____

Address: _____

Occasion (if desired):

- Birthday
- Holiday
- Wedding
- Thank You
- Anniversary
- Other

Memorial Gift:

In memory of (name of deceased)

Send acknowledgment to:

Name: _____

Address: _____

Amount Enclosed: _____

- Check Enclosed
- VISA
- MasterCard

Card # _____

Exp. Date _____

Signature _____

Gift Given By:

Name: _____

Address: _____



Letters to the Editor:

Dear CMTA,

I am a Florida-based, licensed psychotherapist. One of my clients has CMT and was kind enough to put me on your mailing list. I share each newsletter with my clients who suffer from neuropathy. They tell me that they were not aware of many of the resources you suggest in your newsletter. So, I thank you for all my clients who are suffering. —Sue Scott, LSCW

Dear CMTA,

I am 90 years old and use a 3-wheel walker to get around. I pray and hope that a cure is soon found for CMT. I have children and grandchildren and hope that CMT never strikes them. I enjoy your newsletters and like to read about the progress that is being made. —L.L.

Dear CMTA,

I would like to thank you for your newsletter and the quick response I got to a request for information. I couldn't believe that I received your packet with the sample newsletter in less than a week. The thing I appreciated most about the newsletter was the section of letters in which people talked about aspects of their CMT. For someone like myself, who really doesn't know anyone else with CMT, it's reassuring to know that other people deal with the disease much the way I do.

If I can make a suggestion, it would be to increase the articles about practical ways of handling the disease; like articles on braces, physical therapy and such. —J. C.

Ask the Doctor

Dear Doctor,

My 46 year old son has CMT. He has had numerous attacks of kidney stones. He has been told that his body is processing the calcium in his system into the development of these stones and little into bone development. His concern is whether this is a normal condition in patients with CMT. He has experienced fractures in a foot and ankle. Another concern is that he has been told that skin cancer is more prevalent in CMT patients. Is there any truth to this?

He lives in Washington and is having trouble finding a knowledgeable doctor for CMT.

The doctor replies:

I have never heard of any relationship between CMT and kidney stones, nor skin cancer. I suggest that your son contact Dr. Thomas Bird in Seattle, Washington.

Dear Doctor:

I am a 71-year-old man, diagnosed with CMT 1A. One of the physical things that I consider a problem is the apparent lack of moisture in my hands. Added to the usual problems of grasping things with CMT hands, the lack of moisture makes

picking up or holding a glossy or hard-surfaced item difficult. I was told that the lack of moisture in my hands is associated with my CMT. Is that true?

The Doctor replies:

The axons that are responsible for making sweat glands work are not myelinated. Because CMT1A mainly affects myelinated axons, I do not think that your dry hands are a direct consequence of your CMT.

Dear Doctor:

In the last issue of the newsletter, there was a letter from D.S. in NY about tremors. I also have CMT and have a problem with tremors. Are they part of CMT? I have been treated by an ENT doctor for tremors and he prescribed Sinemet to help control them. I also have hypoglycemia that accentuates the tremors when I have not eaten properly. Is this normal?

The Doctor replies:

There are many kinds of tremors. The kind that is most likely to be worsened by hypoglycemia is called "benign essential tremor." It does not have anything to do with CMT. Beta blockers, but not Sinemet, may help.



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.

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, 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*



2700 Chestnut Parkway
Chester, PA 19013
1-800-606-CMTA

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