

The CMTA Report

Vol. 16, No. 6 ISBN #1067-0181

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

CMTA BOARD

- Ann Lee Beyer**
Chairman and President
- Phyllis Sanders**
Vice Chairman
- Richard L. Sharpe**
Treasurer and Secretary
- Robert E. Lovelace, MD**
Chairman, Medical Advisory Board
- Ardith Fetterolf**
Robert Kleinman
Steve O'Donnell
Jack Walfish

CMTA STAFF

- Vincent Bertolino**
Executive Director
- Patricia L. Dreibelbis**
Editor in Chief
- Dotty Lilley**
Program Coordinator
- Edna Rowdon**
Accounting Supervisor

EMAIL CMTA AT:
CMTAssoc@aol.com

The New Look of the CMTA

By VINCENT BERTOLINO, Executive Director

Welcome to the new face of the CMTA! You asked for it and we heard you: the Forum is back. As of this writing 261 people concerned about CMT have signed on. Joining is free and the collective wisdom of participants who post is available for your reading. We hope you will make regular use of this valuable resource and make your own responsible and sensible contributions to the Forum.

A new feature you will see is the availability of shopping. The CMTA has partnered with www.igive.com and www.greatergood.com to provide shopping links to major vendors who pledge part of the sale to the CMTA.

There are hundreds of stores to choose from, including names like Amazon.com, Avon, Baby Gap, Best Buy, Dell Computers, Disney, and FTD Florists, to name a few.

HOLIDAY SHOPPING: BUY ONCE, GIVE TWICE

The holidays are already upon us and if you are like me, you still haven't done your shopping! Well, here is an opportunity to buy gifts for the special people in your life, and, give a gift to the CMTA. When you buy through www.igive.com or www.greatergood.com, up to 26% of what you spend will be sent to the CMTA. My particular favorite is www.igive.com because it tells me how much I have contributed to the CMTA

through my purchases right on the home page when I log in.

PAINLESS FUNDRAISING

You may have thought to yourself that you would like to do more for CMT research, but don't have the time to go raising funds, nor are you or your friends independently wealthy. Besides, it can feel weird asking your friends to send a check. What if you, your family, and your friends could benefit CMT research and

patient services without spending a nickel more than you spend shopping anyway?

Reading this right now, you may be thinking about how much you currently shop online, and how doing so through the CMTA website will do so much to contribute. Now ask yourself, whom do I know who also shops online? Why wouldn't they want to help? It won't cost

them anything they aren't spending now. In my own case, I've given over \$250 in just two months of shopping and I haven't even touched the holiday gifts. Now I'm getting my wife into the site. Funding a cure is just around the corner once she starts hitting the "shop" button!

TREAT YOURSELF—FEEL DOUBLY GOOD

Obviously, the holidays are a big shopping season, but this feature will be available all year long. When you recognize that you need some-

(continued on page 4)



*The CMTA....
Living our
Mission*

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

Vision Statement:
A world without CMT.

Projects and "Wishes"

By **VINCENT BERTOLINO, Executive Director**

We at the CMTA wish happy holidays to you and your family. Your support has helped us provide services for many people with CMT, as well as place new investigators into CMT research. We hope you will continue the tradition of donating to the CMTA this holiday season.

There are many things that need to be done, and these projects may not happen at all without your gift. Let me highlight some of the programs that are in the works:

RESEARCH

Gene regulation—It has been known for some time that the effects of genes can be adjusted, or regulated, to be more or less active. Inappropriate levels of activity cause some forms of CMT. Developing methods of fine-tuning this activity

may yield a therapy. No investigation has been done on this for CMT, yet. Again, your gift is essential for this project to become a reality.

Fellowship programs—The CMTA has supported many fellowships over the years through your generous contributions. To make significant inroads in understanding, there must be more investigators studying CMT. Expanding this program will continue to direct more new scientists into CMT research. Less than 600 scientists worldwide have published on CMT, whereas other disorders have thousands of investigators committed to research. Every gift can make the difference in putting one more scientist on the trail for a cure.

Orthotics and physical therapy—For milder cases of CMT, all that may be needed is to build a better shoe, or determine what modes of physical therapy have the best results in CMT. There are all kinds of new materials that may be effective in making better assistive devices and orthotics. There is also much to do in understanding the action and level of physical therapy most beneficial in CMT. Much of what is done here is the result of isolated physicians tinkering with ideas. Imagine what could happen if we could fund a systematic development program through your gifts.

Not everything is research. Maintaining a professional organization with patient services, and keeping medical research focused on CMT requires projects and programs that also need funding. You might even consider that some of these items belong in the "Research" realm, but they are "Operations" and are critical components to effectively serving the CMT community, both patient and researcher.

OPERATIONS

Promoting the North American CMT Database—Two things need to happen to make this ongoing research project a self-funding success: 1) promotion of free patient enrollment and participation and 2) promotion of investigator utilization. Use of the database is currently free to researchers, but with enough growth in the number of participants, the database will be able to charge a utilization fee and become self-funding. This requires that the CMTA promote the database to patients, physicians, and researchers. Currently, there is little funding for

Conferences such as the recent one held at Wayne State University (see page 10) help connect patients, researchers and other professionals who share an interest in CMT. Shown here at the September 22nd conference, orthotist Bill Messer demonstrates the flexibility of a small orthotic designed to help someone who still has some muscle strength in his or her calf.



doing this. Every dollar you contribute can result in two to three letters to investigators telling them about the availability of this database for research.

Surveys—No other organization focused on CMT has done so much to serve the CMT patient and research community. Most of what our organization does is new, and therefore “uncharted.” There is no guidebook on how to serve the needs of persons with CMT. Through surveys, the CMTA can collect data and analyze it so that the services and overall direction of the organization are in sync with your wishes. We have some funding for this, but we need more. Incidentally, I have included a survey. I encourage you to take the time to fill it out.

New publications—It is not enough to leave research in the lab. New information needs to be added to the medical and non-medical knowledge base. This is done through new publications and periodicals. We also need to reprint old publications with upgraded information, and provide them in other languages. Such resources include a newsletter focused on children, pamphlets on orthotics and physical therapy, and a new and updated physician’s handbook. All of these publications require funding. Getting these projects off the ground won’t take much if many people help with their gifts.

New and upgraded services—The volume of inquiries continues to grow at the CMTA office. While the new website may absorb some of the burden of supplying information and taking literature requests, some people still need a live person to help them. Volunteers can take some of the calls at the support group level, or at the CMTA office, but there is a growing need to have professionals available. One of the ways to address this need is to employ medical and counseling students in university work programs. This provides professional expertise to our callers and creates more CMT advocates in the healthcare and social service sectors. In fact, you may have called us with questions about CMT in the past. What would you have done if we had not been there to answer the phone? We need funding to make these improvements possible.

Thank you in advance for your gift this holiday season. Keep an eye on events in the newsletter; you’ll be pleased with the results we will achieve because you cared enough to help!

CMTA MEMBERSHIP/ORDER FORM

Name: _____

Address: _____

Phone Number: _____ Email: _____

Members who are current with their dues are considered “active.”
If you are unsure as to whether you are current with your member dues, please call the office at 1-800-606-CMTA.

	QTY	COST	TOTAL
Charcot-Marie-Tooth Disorders: A Handbook for Primary Care Physicians		active members \$15 inactive members \$20	
Membership Dues		\$35	
SPECIAL OFFER FOR ACTIVE MEMBERS ONLY: CMT Facts Series (I-IV)		active members \$16	
CMT Facts I <input type="checkbox"/> English <input type="checkbox"/> Spanish		active members \$3 inactive members \$5	
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>			
TOTAL			

Check payable to the CMTA (US Residents only).
Foreign residents, please use a credit card or International Money Order.

VISA MasterCard American Express

Card Number _____ Expiration Date _____

Signature _____

Mail to the CMTA, 2700 Chestnut Parkway, Chester, PA 19013

A copy of the official registration and financial information may be obtained from the Pennsylvania Department of State by calling, toll-free, within Pennsylvania, 1-800-732-0999. Registration does not imply endorsement.

The Annual Board Meeting— Unity in Purpose

Right: New Board Member Steve O'Donnell and Treasurer and Secretary Dick Sharpe discussed fundraising issues for the CMTA.

Below: Chairman and President Ann Lee Beyer endorsed a new vision for the CMTA of "a world without CMT."



Our annual board meeting, held on October 5th, was unique in a number of ways. It was the first board meeting after the September 11th tragedy, and there was apprehension about traveling and what the future would bring. Nevertheless, we had a two-thirds quorum and were able to proceed with business. In spite of recent events, there was an air of optimism and cooperation. As each item came before the board, debate proceeded, motions were made and seconded, and decisions were enacted unanimously.

Much of our efforts was focused on laying the groundwork for a structure that would ensure success in funding the cures for CMT.

This included establishment of a mission and vision statement, by-law revisions, approval of proposed administrative changes, and an open discussion about working with support groups.

We also re-elected two board members whose terms had ended. Carol Henderson was re-elected, but has asked to be excused from the board due to health reasons. Ardith Fetterholf, the former President of the CMTA, was also re-elected to the board.



Board membership is a responsibility that requires active participation and concern for the future of the organization, as well as a willingness to share one's talents and gifts for the good of the group. All present and past members have met these criteria, but departure from the board does not mean that they have suddenly failed to measure up. In fact, board alumni continue to participate

in CMT issues. Dr. Quinn, for example, has resigned from the board after six years, since he needs to spend more time in his practice and with his family. His passion for CMT has not abated, but he recognizes that there are many obligations in his life. He still responds to requests for advice and assistance on CMT, and we look forward to continuing to work with Dr. Quinn as a board alumnus.

We are now a small board of eight dedicated people. We have combined several executive posts, since we no longer need so many chiefs among the braves. As the CMTA grows, we will also grow, not so much in the number of board members, but in the quality and quantity of talent we select to the board. We have a vision—a world without CMT—and we can make it together.

OUR NEW LOOK

(Continued from page 1)

thing, and you're launching your browser to go looking for it on the web, start your shopping through the CMTA site. In fact, you can make the CMTA site your home page if you use Microsoft's Internet Explorer. Just go to "Tools," "Internet options," and click the "General" tab. There you can enter www.charcot-marie-tooth.org and every time you launch, the site will be there.

THAT'S NOT ALL, FOLKS!

Now I've spent a lot of time talking about the ways to shop through our website, but there are

more features, and these are just as exciting. We have added a news archive. There isn't much there, but we hope to change that as we raise the awareness of medical professionals and the general public about CMT. We have an "Ask the Experts" area where you can ask questions which will be forwarded to the appropriate medical professional. As questions accumulate that are relevant to a broad section of the CMT population, we will add these to the list. We have improved the publications section so that you can see what the items look like and how much you save as a CMTA member.

Please take a look around our new site. Tell us what you think. Make some new friends on the Forum, and don't forget to shop!

Thanks for your support!

Pedorthic Management of Charcot-Marie-Tooth Disease

By **MICHAEL LUKOWSKY, C Ped, OST**

Charcot-Marie-Tooth (CMT) disease is a hereditary disorder with progressive peripheral motor neuropathy. This neuropathy leads to progressive distal and anterior atrophy, especially in the feet and ankles. Predominantly, the distal leg muscles and the peroneal and anterior tibial muscles of the foot and ankle are affected, causing progressive weakness with gait disturbances (clumsiness, frequent falls, and tripping over feet). As CMT progresses, the deformity of pes cavus (high arch) worsens and eventually foot drop occurs.

In the early stages, patients begin to have difficulty finding footwear because of the lack of dorsiflexion, due to the atrophy of the anterior tibial compartment. Women have difficulty because of societal pressures to wear dressier footwear, which are typically slip-on styles. They are unable to hold this type of footwear on the dorsum of the foot. Today there are many choices of footwear that have straps or ties that are acceptable as dress footwear.

As the feet of the patients with CMT become more pes cavus, the soft tissue of the plantar surface (sole of the foot) becomes abnormally short. The metatarsal (bones of the arch) heads are lower in relation to the hindfoot at the tarsometatarsal joints. This foot deformity becomes rigid and lacks the ability to absorb shock. Unfortunately, due to the lack of knowledge of the effects of CMT, patients tend to seek out and are guided toward very cushioned and flexible footwear. This type of footwear actually causes more instability to a cavus foot. These patients, in fact, need footwear that has strong rearfoot control with lateral stability. Cushioned orthoses provide the needed shock absorption and support for the pes cavus foot. Intrinsic metatarsal support unloads the metatarsal heads. The combination of stable footwear and cushioned orthoses gives the patient optimum support and comfort.

Due to the high medial longitudinal arch, the extensor tendons and ligaments are shortened, causing dorsiflexion of the distal phalanges (i.e., hammer and claw toes). Therefore, these patients need shoes with deep toe boxes. In today's pedorthic facilities, there are many choices of shoes with deep toe boxes and the new soft stretch toe boxes work well.

As the peroneal (outer) and anterior (front shin area) tibial muscles atrophy, a drop foot develops. Shoes with mild rocker soles are very helpful in reducing toe drag. For a more severe drop foot, thicker, more rigid rocker soles can be attached to most footwear today. In cases where a rocker sole still does not provide enough dorsiflexion (turning of the foot or toes upward) for zero toe drag, leather toe tips are added to the soles to reduce the chance of tripping. Most patients are receptive to trying footwear modifications before an Ankle Foot Orthosis (AFO). In severe cases of drop foot, an AFO is required, but performance of an AFO is enhanced with proper footwear, and rocker soles may still be required. New products like Springlites (thin carbon fiber plates) alter flexion of shoes and improve ambulation and energy return, especially when incorporated into foot orthoses of AFOs.

With awareness of CMT increasing, many physicians are discovering that pedorthic modalities can truly enhance their patients' lives by improving the simple task of walking that most of us take for granted.

Michael Lukowsky is a certified pedorthist at Comfort Shoe Specialists, Inc.

SOURCES FOR HELPFUL INFORMATION

- ◆ www.its-possible.com lists aids for daily living and other issues for disabled persons. There is a downloadable product resource guide available.
- ◆ www.disabilitypreparedness.org offers information on emergency planning and evacuation issues for people with disabilities. This is particularly interesting in view of the Twin Towers disaster.
- ◆ **National Society of Genetic Counselors** can help you locate a genetic counselor. To find a genetic counselor near you: http://www.members.aol.com/nsgcweb/nsgc_hhome.htm or email NSGC@aol.com.



OF INTEREST

Pad your utensils for a comfortable grip. This foam padding allows you to grip smaller items by inserting your forks or other self-help aids in the foam. Order from Sammons-Preston at 1-800-323-5547. Cost is slightly over \$5.



Phenotype Variability in HNPP Simplified

By **MAUREEN HORTON, RN,**
 Edited by **GARETH PARRY, MD**

Currently, in 70% of cases, HNPP has only one genotype or gene problem. This is the deletion of a segment of chromosome 17p11.2-12. This segment of the chromosome contains an important myelin gene, peripheral myelin protein-22 gene (PMP22). CMT1A is genetically a mirror image, containing an extra copy of PMP22 at the same site that HNPP has a deletion. This one gene deletion, which causes HNPP, has different patterns of presentation.

Phenotype variability is basically a way of saying there are different ways that HNPP can look and act. These are the symptoms a doctor may see when a patient comes into the office. It is important for doctors to recognize the many different ways HNPP may manifest itself. Dr. Gareth Parry says, “We have tried to focus on the presenting feature of the disease. The rationale is that they need to be diagnosed correctly when they are first seen so we want people to recognize what the disease can look like when it first appears. Thus, the different phenotypes are those that are present at the time of the initial presentation.” Dr. Parry went on to explain that, over time, almost all patients go on to develop other features and they can change from one phenotype to another.

So, if phenotypes are a tool for the doctors, then why would we, as patients, care about them? Actually for several reasons:

- Once they know that different phenotypes exist, people with HNPP are curious to know which type they are.
- Other family members may not have the same phenotype (which is one of the reasons this can be so hard to diagnose). Knowing the different phenotypes is a way to recognize the symptoms, so that those family members, who have not been diagnosed yet, can get diagnosed.
- Doctors are noting that people may switch from one of the five varieties or phenotypes to another. Tracking your own phenotype is a way to watch the progression of the disease. It should be noted, however, that

some people don't switch and have the same phenotype for life.

- And finally, it is an alert to those diagnosed with CMT who may actually have HNPP instead.

The number of phenotypes has changed. For the past several years, five phenotypes have been listed in the description of HNPP: classic HNPP, acute arm paralysis, polyneuropathy resembling CMT (CMT), confluent mononeuropathy multiplex (CMM), and the oligosymptomatic phenotype. The oligosymptomatic phenotype was, by far, the largest group and composed of those who had very few or mild symptoms and who often have not sought medical help. Some reviewers of the medical literature say, and with some justification, that a phenotype describes a clinical appearance and if the patients have no symptoms, they don't have a clinical appearance. Thus the oligosymptomatic group cannot really be a phenotype. In keeping with the new thinking, the oligosymptomatic phenotype will be dropped. But a description of this largest group of HNPP carriers will be included after the four remaining phenotypes are discussed.

With the revised classifications there are currently four major phenotypes: classic HNPP, acute arm paralysis, polyneuropathy resembling CMT (CMT), and confluent mononeuropathy multiplex (CMM). In addition, many patients are asymptomatic or have so few symptoms that their disease is not recognized (previously the oligosymptomatic phenotype). The first two categories, classic HNPP and acute arm paralysis, are characterized by episodic problems. Polyneuropathy resembling CMT (CMT) and confluent mononeuropathy multiplex (CMM) phenotypes are characterized by their more persistent and progressive symptoms that are moderate to severe. Both CMT and CMM phenotypes can have numbness and weakness and other signs of a generalized neuropathy. A very simplified way to distinguish the latter two is that in the CMT-like phenotype symptoms are mostly symmetrical, while in CMM they are not symmetrical. Each phenotype will be discussed in more detail.

AN OVERVIEW OF PRESSURE PALSIES

In order to understand the phenotypes of HNPP, one must first understand what is meant by the term “pressure palsy”. When HNPP was first described by DeJong in 1949, it was called “bulb diggers disease”. DeJong noted the periods of weakness and numbness (palsies) in bulb diggers and thought they were caused by the kneeling position and pressure on the peroneal nerve. Thus the term pressure palsy. The spectrum of this disease has evolved over the past five decades, but the concept of pressure palsies has remained. We do know that the pressure, which DeJong described, can and does cause episodic numbness and weakness. But we also know that the nerves are in general very susceptible to injury and can also be injured by external pressure, stretch, repetitive use, and the build-up of small injuries over time (cumulative effect). We also know that a progressive generalized neuropathy is an integral part of the disease, even in those patients who never recall having a pressure palsy episode (Parry).

Pressure palsies are typically described as transient (or intermittent or episodic), painless and often recurring symptoms of numbness (or tingling) and weakness. (Physicians often use the terms *sensory* and *motor* to describe the symptoms. *Sensory* means the sensations the patient feels, such as tingling, numbness, or pain. And *motor* refers to strength or weakness.) These episodes can be as brief as a few minutes, but can also last several days or even several months. Numbness may be as mild as the individual noticing that an area or limb does not have quite the same feeling as surrounding areas on the other side, or so severe as to feel like the area or limb has been shot full of Novocaine. Weakness also can vary between slight and barely there to so severe that the individual is unable to move a particular muscle group or the entire limb.

The most common problem sites are the wrists with carpal tunnel syndrome; the elbows with cubital tunnel syndrome; and the knees with peroneal nerve injuries. But any peripheral nerve—outside the brain and spinal cord—can be affected. It would be rare to see problems with nerves in the trunk, but fingers, elbows, scalp, shoulders, toes and feet are also frequent trouble spots.

PROGRESSION OF HNPP

As mentioned above, over time, individuals can change from one phenotype to another. This typically involves changing from a milder or episodic form to a more permanent and generalized neuropathy phenotype, such as CMT or CMM.

Although people who carry the HNPP gene deletion may have no symptoms initially, as time goes by, and as people are evaluated more carefully, it has become clear that mildly affected individuals often have symptoms attributed to other common disorders such as carpal tunnel syndrome or lumbar disc disease. Some researchers believe that eventually (and this may take a lifetime), all individuals show signs of a generalized neuropathy. Generalized neuropathy usually means more widespread symptoms of permanent numbness and weakness. The numbness is often a “stocking and glove” distribution and the weakness can cause foot drop and hand weakness. And it may not be until the general neuropathy has developed that the person first sees a physician.

The nerves in general are very susceptible to injury...caused by external pressure, stretch, and repetitive use....

For some the HNPP symptoms progress to a generalized neuropathy very slowly. For others the progression is quite rapid. Some are younger, others are older when this happens. Medical science does not yet understand why there is such a wide variance in symptoms, speed of development or age of onset.

There also appears to be a cumulative component to HNPP symptoms and activity. We tend to think of each nerve as one single nerve. In fact, there are many nerve fibers making up the one nerve. Some people report being able to do an activity one day without any problems, but have increasing symptoms if they continue to do the same activity many days in a row. When people experience an increasing problem such as this, they are thought to be damaging more and more of the nerve fibers. Although nerve fibers are probably damaged on the first day of the activity, there are enough fibers still functioning to compensate for the damaged ones. As the activity continues, more and more fibers are damaged and the symptoms become more apparent as the nerves are unable to continue to compensate for the injury. This cumulative component of HNPP can make pacing activities extremely difficult for individuals with frequent HNPP symptoms.

(continued on page 8)



VISIT OUR HOMEPAGE

at

www.charcot-marie-tooth.org

How to Find The Information You

Adapted from Generations, a publication of the National Ataxia Foundation

People dealing with disabilities often need specialized information in order to participate fully in life's activities. Often it's difficult to figure out where to find this information or even what questions to ask. The issues raised by a disability affect so many aspects of life that the range of possible solutions together with the rapid changes being made today present a daunting challenge for people seeking useful information.

Information can be obtained from many sources, such as knowledgeable individuals, agencies, printed materials, the Internet, or meetings and conferences. At some point, you might choose to meet with people who have lived with the same situation as you in order to learn how they have managed. Their solutions may not be suitable for you, but you will get an idea of the options and alternatives that are out there for you.

Some possible topics for your searches might include issues surrounding education, employment, access and accommodations, the law and government programs, assistive technology and even details about the disability itself. No one person or organization will have all the answers, so persistence is necessary.

WHERE TO LOOK

Telephone Book—Your telephone book is, in fact, a resource, not just the white pages but the special sections which list community and government services.

Human Services Yellow Pages—This special “telephone” book which lists human service agencies, private organizations, and other groups



dedicated to meeting the special needs of people living with disability, should be available at your local library.

Government—All states and many cities have at least one office dedicated to the needs of people with disabilities. These include the Office on Disability, Rehabilitation Commission, Commission for the Blind, the Commission on Mental Health, and others. Call any one of them and ask them to refer you to the resource you need.

Independent-Living Centers (ILCs)—ILCs are essentially self-help centers that are run and staffed by persons with disabilities. They offer some or all of the following services: attendant care, coordination of personal care attendants, transportation, counseling, information and referral, independent-living skills training, and self-empowerment. Most important, you will find people who have faced and understand questions very much like those you are dealing with.

Hospitals and HMOs—Recently the health care organizations have begun to take a more active role in many more areas of health. Your local hospital or HMO is likely to have a great deal of information on disability, hot lines for information, “talk to a nurse” programs, and a wide range of support groups. Call your provider first and then call every health care provider in your area until you find programs and services that meet your needs.

Disability-Specific Organizations—Most major disability-related organizations focus on one disability, or perhaps several related disabilities.

HNPP

(Continued from page 7)

Because HNPP is primarily a demyelinating neuropathy, recovery from palsy episodes is usually complete at first, with numbness and weakness going away entirely as the nerves remyelinate. So, at first, the nerves are damaged and eventually heal completely. Over time, with repeated injury, the nerves are damaged and only partially heal. And with further injury, the damaged nerves again only partially heal. So the

progression of the disease is typically seen as a very slow step-down pattern, with times of no progression, followed by progression. For some individuals, there may be years between pressure palsy episodes. Others have mild problems for years and then begin to progress at a more rapid rate. In rare instances, some individuals develop their first symptoms and remain on a fairly steady downward course.

In the next issue we will begin examining each of the phenotypes in detail.

Need

These national organizations, such as the CMTA, usually have local support groups which you can contact. They have publications, meetings, and conferences which can provide both resources and networking opportunities, many concentrated primarily on research and education.

Work—Because of the passage of the federal Americans with Disabilities Act, most employers have assigned someone to be the ADA Compliance Officer. This person can answer many disability-related questions and access or refer you to resources. If your or a family member's disability is affecting your job, talk to your human resource office.

Library—Your library and the librarian are resources. They have books on disability, resource guides, publications of disability organizations and effective catalogs to help you find them. In addition, today, many libraries can give you access to the Internet.

Schools—With new laws and growing awareness, the school has become a disability resource. The school nurse, guidance counselors and special education staff can offer information and referrals. Colleges have centers and programs for students with disabilities. You do not have to be a student or have a student in the school to seek their advice.

Internet—The Internet not only has a wealth of disability-related information, it may have too much. If you search the Internet with the word “disability”, it will take hours just to read the titles of the sites you will find. However, if you have a good browser and ask specific questions, you can find unlimited useful resources. One of the nice things about the Internet today is the number of people who love to spend their time “surfing.” Feel free to ask even a casual friend who is “into” the net to search for you; more likely than not he or she will be very happy to have a mission to justify the time he or she spends “on the net.”

Support Groups—There are hundreds of support groups that focus on specific disabilities. They are made up of people just like you: persons with disabilities and their family members. They often have local meetings where people gather to share experiences and information. They can give you the opportunity to ask questions and share solutions. You can find them through disability-specific organizations, the community service events section of the newspaper, a local hospital, or the Human Services Yellow Pages.

UPDATE FROM THE CAPITAL

Rare Diseases Act Introduced

(Because of the World Trade Center disaster, most bills of this sort are on the back burner for the time being.)

On August 3, Senator Edward Kennedy and Senator Orrin Hatch introduced the Rare Diseases Act of 2001 (S. 1379). Not since the passage of the Orphan Drug Act has such an important piece of legislation been introduced that positively impacts the entire rare-disease community of 25 million Americans.



In the text of S. 1379, Congress made the following findings:

For many years, the 25 million Americans suffering from over 6,000 rare disorders were denied access to effective medicines because prescription drug manufacturers could rarely make a profit from marketing drugs for such small groups of patients.

The Orphan Drug Act (ODA) created financial incentives for research and development of such “orphan drugs”.

Despite the tremendous success of the ODA, rare diseases and disorders deserve greater emphasis in the national biomedical research enterprise.

The National Institutes of Health (NIH) has received substantial increases in research funding from Congress for the purpose of expanding the national investment in behavioral and biomedical research. Notwithstanding such increases, funding for rare diseases and disorders at the NIH has not increased appreciably.

The Food and Drug Administration (FDA) supports small clinical trials on new treatments for rare disorders through Orphan Products Research Grants. Yet, the appropriations in FY 2001 for such research grants were less than in FY 1995.

What does the Rare Diseases Act of 2001 do?

1. Provides a statutory authorization for the existing Office of Rare Diseases (ORD) at the NIH.
2. Increases the national investment in the development of diagnostics and treatments for patients with rare disorders.
3. Authorizes regional centers of excellence for rare disease research and training.
4. Increases funding for the NIH Office for Rare Diseases to \$24 million for fiscal year 2002 and “such sums as may be necessary for each subsequent fiscal year”.
5. Increases the funding for the FDA's Orphan Product Research Grants program, which has provided vital support for clinical research on new treatments for rare disorders to \$25 million for fiscal year 2002, and “such sums as may be necessary for each subsequent fiscal year”.

CONTINUING OUR PATIENT-EDUCATION PROGRAM:

A Conference at Wayne State

Despite the tragic events of September 11, 2001, 65 people braved traveling to attend the patient-family conference on September 22 in Detroit, Michigan. The conference was the second that has been held at Wayne State University's Harper Hospital. The proceedings were organized by Dr. Michael Shy and Karen Krajewski, both of whom were general session speakers.

The actual weekend events began on Friday night, September 21, 2001, with an open house and tour of the research facilities at Wayne State, hosted by Dr. Shy. He personally led the tours and answered questions of the people who were able to attend. Although the turnout Friday evening was small, the people who were present had the pleasure of having their individual questions answered and their problems assessed by Dr. Shy and Karen Krajewski. In addition, Raven Lewis, a staff member of the CMT Research Team, was present to answer questions about the CMT North American Database.

On Saturday morning, the conference began with registration and a continental breakfast. In the first presentation of the day, Dr. Shy gave an overview of what is known about CMT and what directions research is taking. He discussed the need to identify new genes, understand the pathogenic mechanisms of the disorders, and improve pre-implantation technology and gene therapy. Karen Krajewski, MS, followed with a

Attendee Paul Arthur had Dr. Shy's undivided attention during a question session at Friday night's open house.



Robin Goodrich, a regional representative from Athena Diagnostics, explains the various DNA tests currently available to diagnose CMT.

discussion of the new genetics booklet and genetic counseling issues. She then talked briefly about the CMT North American Database and the need for more families to complete and return the surveys.

Following a short break, the first round of workshops began. One on pain management was led by Medical Advisory Board member, Dr. Richard Lewis, another on genetic issues was hosted by Karen Krajewski, and the final workshop on rehabilitation issues was co-directed by Dr. Shy and orthotist Bill Messer.

After a break for lunch, there was a final workshop on the medical services offered by Athena Diagnostics, Shriners' Hospitals, the Wayne State CMT Clinic and the CMTA.

The final presentation of the day was by Dr. Barbara Kamholz on the stress of dealing with a chronic condition. Dr. Kamholz is a clinical psychiatrist whose insight into the stress of disease as well as the stress of being a caregiver was helpful. One of the points she emphasized was how a sense of loss contributes to our feeling of

Nantahala Neuropathy

By **JAMES LEWIS, MD, Franklin Neurology Clinic, Franklin, NC**

Last year the CMTA was kind enough to run a query in *The CMTA Report* concerning CMT and trigeminal neuralgia. I thought the readers would like some follow-up.

Initially, I saw a patient with severe episodes of facial pain known as trigeminal neuralgia (TN) and a sensory polyneuropathy. She had pes cavus. Her mother had a numb face, sensory neuropathy and pes cavus. There was a strong family history of similar findings in earlier generations. I found literature references to families with TN and CMT and became interested in the problem. One of my first steps was to place the request for contacts with similar patients in this newsletter. I got 13 appropriate responses (and a few that were completely unrelated to the issue). In 11, there was no family history of TN; in two, individuals reported multiple generations with TN and CMT. One patient spoke with me once on the phone, but did not keep in touch. The other lived in Oregon; my family was in North Carolina. However, we quickly found that she was a descendent

of my family whose grandmother had moved West years before. That was a Twilight Zone moment for both of us!

At this point, I have fully evaluated six people from this family who all have CMT1 on the basis of NCVs. Five have a predominant sensory neuropathy, with little to mild weakness. One patient had the explosive onset of severe weakness and wasting in his legs in his 50's, after years of only the mildest possible problems with his ankles turning over. Three have TN, three have twitching movements of the face (hemifacial spasm or HFS), and four have hearing loss. They all have a previously unreported mutation of the MPZ gene, making this an unusual variant of CMT1B. In two unaffected relatives, the mutation is absent; and two unrelated people with a family history of TN are also negative for the mutation. All the patients have a poor sense of smell, most have severe restless legs and many have migraine. This clinical picture is so unique that, while recognizing that the family clearly belongs in the group of CMT disorders, I have suggested that their variant deserves a special name: Nantahala neuropathy, after the Indian name for the region of North Carolina where 10 generations of the family have lived.

I presented a poster of this information at the recent World Congress of Neurology. One of the numerous interesting oddities of this story is that some family members were reported by the Cleveland Clinic in 1977. It was recognized as a distinctive picture at the time; but this was before genetic testing was possible, and the unique status of the disorder could not be determined. My patient with the late onset of wasting was seen in Cleveland in his 30's, and was noted to have abnormal NCV's, but no clinical findings.

On the basis of the responses to my survey, it seems to me that most types of CMT can be present with isolated cases of TN at about the same rate as the rest of the population—about 1 per 1000 individuals. The Nantahala variant is unique in the extent of cranial nerve involvement; why TN, HFS, decreased smell and hearing loss are so common has yet to be established.

My coworkers on this study were William Allen, a geneticist at the Fullerton Genetics Clinic in Asheville, NC; Christy Barbee, a student at Western Carolina University; and William Seltzer, the medical director for Athena Diagnostics. A manuscript is being prepared for submission to a neurology journal.

DETROIT CONFERENCE

(Continued from page 10)

hopelessness. For someone with a chronic illness, the sense of loss involves a loss of independence, a loss of self-sufficiency, a loss of health and a loss of self-image. In addition, chronic pain predisposes a person to depression, which makes a person function worse. But Dr. Kamholz was definitive in saying that every single thing you can fix helps your overall equilibrium, and depression is one thing that can be fixed!

Caregivers also have higher rates of depression than the general population—up to two times higher, according to Dr. Kamholz. If caregivers believe that care is a burden, they are more likely to suffer their own illnesses, both emotional and physical. Following her presentation, Dr. Kamholz fielded a number of questions about children and CMT and the sense of guilt that surrounds passing on a genetic disorder.

The conference ended with an open question-and-answer period for the entire group of attendees and presenters.

Disability and Quality of Life in

By G. PFEIFFER, E. M. WICKLEIN, T. RATUŠINSKI,
L. SCHMITT, K. KUNZE

Editor's note: This article is the source of the comment on the new home page that CMT Type IA has a severity comparable to that caused by stroke. Because of discussion on that statement, we present the source paper for our readers. It is reprinted from The Journal of Neurology, Neurosurgery and Psychiatry 2001;70:548-550.

ABSTRACT

Objectives—Charcot-Marie-Tooth disease type 1 (CMT1) is a hereditary sensorimotor neuropathy causing variable degrees of handicap. The risk for relevant disability in respect to genetic counseling is unknown. An attempt was made to define it.

Methods—Disability and ambulation of 50 patients with CMT1 were scored by the Hauser ambulation index score and the Rankin scale. Rankin score 2 was subdivided into 2a (independent without relevant slowness) and 2b (independent, though at the cost of excessive time consumption). The sickness impact profile was assessed and compared with patients 6 months after stroke who were without mental deficit. To define at which degree sickness and disability become relevant for genetic counseling, the patients were asked whether they would refrain from childbearing if the children were at risk of inheriting a disease that caused as much disability as they experienced themselves.

Results—Subdivision of Rankin score 2 was reliable and improved validity. High disability significantly predicted an attitude against childbearing (stepwise logistic regression) only with this subdivision. Thirty-six percent of the patients voted against childbearing. The cut-off for relevant disability in respect to childbearing was a Rankin score higher than 2a, which was present in 44% of the patients. Psychosocial impact was comparable with patients with stroke and similar disability. Depression was present in 18% of the patients.

Conclusion—Subdivision of Rankin score 2 is recommended for the assessment of longstanding disability in neuromuscular disorders. Disability becomes relevant for the attitude towards childbearing as soon as everyday activities become markedly slow (Rankin score 2b). Relevant disability occurred in 44% of the patients. Emotional stress in CMT is similar to that of patients with stroke and comparable disability.

Prenatal diagnosis for Charcot-Marie-Tooth disease type 1 (CMT1) is now available. For adequate counseling, we have to know more about the impact of the disease. Information about the inheritance risk may be sufficient in uniformly

severe disorders such as Duchenne muscular dystrophy. Even then, prospective parents ask for the consequences of the disease. Charcot-Marie-Tooth disease type 1 has been designated as a relatively mild disease. Others say that 20% of the patients are seriously handicapped. This information is too contradictory. Disability has not yet been properly assessed in CMT1. Use of the neurological disability score measures impairment rather than disability. Only 60 of 104 patients with CMT were able to assign themselves a score between 0 (no effect) and 10 (every activity impaired). Other studies did not consider the arms, which are often involved in CMT1. To our surprise, the Rankin scale, a standard measure of disability in stroke, multifocal motor neuropathy, and CIDP, has not yet been used in CMT1.

Our patients often realized their disability only after queries about professional and past-time activities, or when we witnessed their slow undressing. Timed motor activities in CMT1 are up to six-fold prolonged. Therefore, we subclassified Rankin score 2 for independent but exceedingly slow patients. To illustrate the burden of CMT1, we also compared it with a more frequent and better known disease: stroke with predominantly physical disability.

We asked our patients whether they would advise against childbearing, if the prospective child would have similar disability. In this hypothetical setting, a vote against childbearing implies that the patient considers the disease to be so severe that it is better not to start a life with the disease. It, thus, informs about perceived quality of life, and defines "relevant disability" in regard to prenatal diagnosis.

PATIENTS AND METHODS

Methods—The study was approved by the local ethics committee. A structured interview explored time consumption and disabilities for professional and past-time activities. The Rankin scale was prospectively modified by sub-classification of independent patients who required more than twice the time for everyday activities than healthy companions or colleagues (Rankin score 2b). Two independent observers (E W and T R) concurred in this respect in 31 of the 33 patients. Ambulation was scored according to Hauser et al. Disease impact was measured by a German translation of the sickness impact profile (SIP) validated for musculoskeletal disorders. The SIP provides 141 questions addressing

Charcot-Marie-Tooth Disease Type 1

problems in 12 categories of everyday functions. Affirmative answers are weighted and summed up for every category. The scores are reported as the percentage of the maximum impact. We used the self rating depression scale (SDS).

Twelve biographical, physical, and psychosocial variables were related to the attitude towards childbearing by O2 (nominal items) or Mann-Whitney tests (ranked scales) and by logistic regression (SPSS for Windows). Seven patients were excluded from multi-variate analysis because of missing values. Variables were selected stepwise backwards, using the conditional statistic. Logistic regression models are linear combinations of the independent variables and yield Z scores which allow the calculation of how probable it is that a given person has a positive or negative attitude towards childbearing. Comparison with the actual attitude of the patients shows how good the model explains the attitude.

Patients—Fifty patients met clinical criteria for CMT1; 35 index patients and 15 affected relatives did not differ by Rankin score ($p=0.32$). Forty-six patients had a positive family history. The four sporadic patients had a duplication on chromosome 17p11.2, which was shown by Southern blot hybridisation in 29 patients. Neither patients nor partners were pregnant. Twelve patients had unaffected children. At least one child had CMT1 in 12 families. The SIP was determined at the 6 month follow-up of 23 successive patients with stroke without aphasia or mental deficit according to the mini mental state examination.

Results—Disability neither depended on age ($p=0.60$), nor on the presence of a duplication. Gait was normal in five patients (Hauser ambulation index score (HAS) 0 or 1). Twenty-seven patients with abnormal gait managed 8 meters in less than 10 seconds (HAS 2). Fifteen patients walked without support but required between 10 and 20 seconds for 8 meters (HAS 3). Manual dexterity was impaired in 26 patients. Twenty-seven patients had deteriorated during the past 5 years. The disease influenced choice of profession or necessitated re-training or early retirement in 36% of the patients. Related to the Rankin score ($p=0.02$), the disease interfered with professional life in 58% of the patients. The 35 patients with restricted past-time activities had higher physical ($p=0.013$) and psychosocial ($p=0.025$) SIP scores. Thirty-four patients complained of neuropathy related pain

or painful muscle cramps. Seventy-six percent of the patients consulted doctors because of neuropathy related complaints; 38% of the patients regularly received physical therapy; 52% used orthopaedic devices. The SDS indicated depression in 8 of 50 patients, unrelated to disability ($p=0.48$).

The median SIP scores were similar to means of elderly control subjects except for “sleep and rest” and “body care and movement”, which exceeded the control means in more than 75% of the patients with CMT. Neither the Rankin scores nor the SIP scores “emotional behavior”, “body care and movement”, “ambulation”, and “eating” differed between patients with CMT1 and those with stroke. The other SIP scores were significantly lower in CMT1. The 42 SIP outliers indicating exceptionally high impact were from 18 patients.

Thirty-two patients favored childbearing. Low Rankin score ($p=0.014$) and the fact that the patient already had children ($p=0.023$) predicted a positive attitude towards childbearing. No patient with Rankin score 1 and a quarter of the patients with Rankin score 2a discouraged childbearing, whereas five of nine patients with Rankin scores 2b and seven of 12 patients with Rankin score 3 did so. The full regression model with all variables predicted the attitude towards childbearing correctly in 81.4% of the patients. The final model:

$$Z = 1.54 - 1.43 \times (\text{Rankin score}) \\ + 2.02 \times (\text{presence of children}) \\ + 2.25 \times (\text{presence of affected family members from the previous generation})$$

predicted the attitude correctly in 76.7% of the patients. The different signs for “Rankin score”, and “presence of children” and “presence of affected family members” indicate effects in opposite directions: high disability predicts a vote against childbearing, whereas presence of own children or affected family members favors childbearing. Various backward selection starting from subsets of the full model selected the same final model. After reclassification of Rankin scores 2a and 2b into 2, disability dropped from the logistic regression model.

Discussion—We expected higher emotional stress in patients with stroke because they have less time to develop successful coping. However, emotional stress was comparable in both dis-

(continued on page 14)

QUALITY OF LIFE

(Continued from page 13)

eases. Patients with CMT scored unexpectedly high in two SIP domains: “sleep and rest” and “alertness behavior”, due to frequent affirmations to “I sit during the day”, “I lie down more often during the day in order to rest”, “I have minor accidents”, or “I react slowly to things that are said or done”. These statements infer something different from that intended by the SIP designers, but illustrate typical problems of our patients, who behaved clumsily, which may provoke teasing already at school age, stamping them early as outsiders who, when thriving at normal professional achievements, will have to fight permanently because they are slow in important activities. Higher rest requirements and pain are additional burdens. The high percentage of patients seeking medical help also falsifies the idea that impairment is hard to perceive in CMT1. The worst case scenario for prospective parents deciding about prenatal diagnosis includes the facts that CMT1 affects choice of profession, severely slows important activities,

requires help in everyday life, and causes emotional distress similar to other chronic conditions.

The split between irrelevant and relevant disability passed through Rankin grade 2: disability dropped from stepwise regression after reclassification of Rankin scores 2a and 2b into 2. We suggest a division of Rankin grade 2 for CMT and other diseases with longstanding physical disability.

Disability was similar to that found in other studies. Forty of 119 patients with CMT1A were unable to run: 56% of our patients had similar walking difficulties (HAS > 2). Another study found a similar degree of ambulation impairment (walking obviously impaired, running impossible) in 42% of their patients. Relevant disability (Rankin score > 2a) was present in 44% of our patients. The vote against childbearing (36%) underestimates the prevalence of relevant disability because family factors opposed its effect, as shown by logistic regression. Therefore, the inheritance risk in mathematical decision models for prenatal diagnosis should be weighted by 0.44, corresponding to the percentage of patients with Rankin grade above 2a.

Foot Care for the “Numb” Foot

By **DONNA CLARK**

I have some suggestions for people who are having trouble with the feeling in their feet. If problems are caused by the foot care you are currently receiving, which means that the shoes and braces that you are wearing rub any sores or calluses on your feet and legs, even if you cannot feel those sores, you need to address that issue! If your body is working to heal a sore, that takes energy from the rest of you. The sore also means that the devices you are wearing are not allowing you to be efficient in your walking. Go to the professionals who fit you and ask for help in reducing the rubbing and sore spots.

Fitting shoes on feet with reduced sensation requires the foot care specialist to have a better knowledge of your foot than you do. The shoes you wear should be big enough so you can wiggle your toes. Some of you can no longer wiggle, so you should draw around your feet while you are sitting, cut out your foot shape and lay the pattern into the bottom of your shoe. If the heel area fits well and you have about 1/2 inch at the ends of your toes and you have all the toes in the shoe and are not crowded, then your shoes should not rub your feet and cause sores. If your shoes do not fit this way, there is a professional out there to help you.

Shoes must look like your feet also. No one has a “high-heeled pointed toe”- shaped foot, so a lot of cramming goes on. Women can get by wearing dress shoes for a 2-hour period, but the feet should be monitored after the 2 hours for red places or rubbed areas.

If you cannot feel your feet well, you can rub a spot and have a bad infection within 3 hours. These pressure infections usually do not get better on their own; they will need a doctor's care. Remember that you will look best if your feet are intact and not rubbed red and sore.

With neuropathy or loss of sensation, a new shoe should be worn for two hours, and then taken off. Socks should be taken off and the feet looked at. If you cannot look at your entire foot, get a mirror that magnifies and look all over. If you have red or rubbed areas, do not put the shoes back on until you have sought help with the shoes.

Another protection you need to employ is checking the shoes each day with your hands before you put your feet into them. If you have trouble with your hands, use a small flashlight and shine it into the shoes. We have one man who wore his grandson's small car in his boots all day and ended up having surgery that evening to remove the car. A lady wore a clothes pin in her dress shoes for 3 days! Fortunately she was in a bridge tournament and did not walk much. I almost had a heart attack when I found it during a check-up.

Remember you do not grow new feet, so take care of what you have, even if they are not pretty.

The Herb of the Month: Ginkgo Biloba

By **BRUCE A. CRISTOL, PharmD**

From the leaves of the Ginkgo tree, also known as the maidenhair tree, kew tree, and yinhsing, ginkgo biloba is derived. The ginkgo tree is considered to be the world's oldest living tree species, dating back more than 200 million years. Although not living as long as the famous Sequoia gigantia of California, ginkgo trees can nevertheless survive as long as 1000 years. Growing to a height of approximately 125 feet. The tree produces fan-shaped leaves. The highest concentration of the active components in the leaves occurs in autumn. Numerous chemicals have been identified within the leaves, including, for example, about 40 different flavonoids (aromatic chemicals), as well as numerous different organic substances.

In China, where ginkgo survived the Ice Age, the tree was cultivated as sacred and it has been used there as a medicinal for more than 1000 years. Traditionally Chinese physicians prescribed ginkgo for asthma and chillblains (swelling of the hands and feet from exposure to damp cold). Both the Chinese and Japanese ate roasted ginkgo seeds to aid digestion and prevent drunkenness. With technologic advances over the past 40 years, specifically

involving the purification and extraction of ginkgo's essential compounds, Europeans have utilized ginkgo as a prescription medication, in both oral and injectable forms. However, in the United States ginkgo is only sold as a nutritional supplement.

Ginkgo has been used for cerebral insufficiency (by improving blood flow), anxiety/stress, tinnitus ("ringing in the ears), circulatory disorders, dementias (such as Alzheimer's), and asthma. Side-effects are uncommon but have included headache, dizziness, heart palpitations, gastrointestinal reactions, and skin eruptions.

Ginkgo biloba is not considered a "drug" by the U.S. Food and Drug Administration (FDA). 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. Drug-herb interactions have been uncovered in the past several years which the patient should be aware of.

Until Ginkgo biloba 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 follows:

Drug interactions with Ginkgo biloba	Result of interaction	Comments
Aspirin	Increased bleeding time*	This interaction may also apply to ibuprofen and naproxen.
Dipyridamole (Persantine®)	Increased bleeding time	
Warfarin (Coumadin®)	Increased bleeding time	
Thiazide diuretics (e.g. Hydrodiuril®)	Hypertension	Hypertension has not been associated with ginkgo biloba alone.

* Aspirin in combination with ginkgo biloba has been associated with hyphema (bleeding into the eye)

REFERENCES:

DerMarderosian, Ara. The Review of Natural Products. *Facts and Comparisons* 1999.

Fugh-Berman, Adriane. Herb-drug interactions. *The Lancet* 2000; 355:134-138.

Miller, Lucinda. Herbal Medicinals: Selected Clinical Considerations Focusing on Known or Potential Drug-Herb Interactions. *Archives of Internal Medicine* 1998; 158:2200-2211.

Support Group Leader Profile: Robert Budde

When my father was in his mid-forties, he noticed he was having problems with his hands and legs. Doctors then never came up with any diagnosis that they could attach to his symptoms. He died at age 75 of a heart attack, never knowing what his diagnosis was.

My older brother also began having signs and symptoms in his hands and legs. He was never one to talk much about his problem but felt it was the same thing our father had.

My first signs of CMT came in my mid-forties. I think I first noticed my problems with my hands and later my legs. It wasn't until 1978 when we moved to Dallas, Texas, that a physician there put the name of Charcot-Marie-Tooth to my condition. Since my father had never let things stop him, I guess I figured I would adjust to my weaknesses as he had done.

In 1993, we moved to Lexington, Kentucky to retire near our two daughters. It was here that I went to the University of Kentucky and confirmed my diagnosis through their neurology clinic and had nerve testing done to determine I had Type 1. Since I was then retired and needed something besides my golf game to keep me busy, I decided to start a CMT support group. My brother and nephew who also have CMT had gone to a CMT meeting in Akron, Ohio and had some information on support groups. I contacted the CMT Association and they sent a mailing out to members in Kentucky, southern Indiana and southern Ohio. I also worked with



Phyllis and Bob Budde work together to keep the support group active and informative for everyone.

the Muscular Dystrophy office here who also sent out mailings for us. Our first meeting was April, 1998 with eight persons with CMT and two spouses. We also had three people call and say they were interested but could not attend.

We now have 26 names of persons with CMT on our mailing list and meet quarterly, inviting speakers on subjects of interest to the membership. We are lucky to have developed a group of people who seem to enjoy meeting and sharing problems and information.

Support Group News

■ Kentucky/Southern Indiana, Southern Ohio

The November 10th meeting involved spirited discussion about the availability of aids for people with disabilities and how to get help from the Central Kentucky Independent Living Association. That group stresses the importance of people being responsible for their own needs. They enable individuals to attain that goal by helping them make changes in themselves and their environment. Independence Place has services involving peer counseling,

information and referral, independent living skills and personal and public advocacy.

■ Minnesota/ Benson

The fall meeting was a huge success according to Rosemary Mills. There were 11 CMT persons in attendance along with five spouses. Four of the attendees were new members. They watched a video on CMT and then shared personal stories of dealing with the disorder. In sharing, the members find a common bond.

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) Burrow,
 336-434-2383

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

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

Ohio—Greenville

Place: 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:

Marjorie Bank

Ken & Cathy Weaver

Herman Dees

Kathy Kersh
Katherine Touchstone

Paul Friedman

Cecle & Joe Adler
Dr. & Mrs. Stephen Brennan
Faith & Morty Brown
Betsy, Phil, Rachel & Sarah Darivoff
Dr. & Mrs. Robert Israel & Family
Patricia Falcone & Dorothy Kreindler
Jen & Bill Stasior
M. L. & Heni Schwartz
TIAA-CREF BIRS Staff
Leslie & Joel Wasserman
Mr. & Mrs. Jack Weinberg

John Hill

Dr. & Mrs. Gerrit Gucky
Shirley Martin

William Lamnick

Gerald & Elsie Belloli
Warren & Barbara Galle
Larry Kimball
Rick, Bonnie Kerrie, Kage Kimball
George & Carol Saufley

Elaine McCarthy

Carol Gartland
Carl & Margaret Hadley
Joan Jones

Mildred Pasquenza

Betty Phillips

Gino Pisaneschi

Kenneth & Diane Bond
Luci Callahan
Anne Camp
Robert Conway
Mr. & Mrs. Lee Eckert
Jean & Gene Fedor
Maryanne Gelso
Carol Harding
Harriet Hutsko
Beth Laerber
Cinde Linker
Christopher Loesch
Teresa McGowan
Ellen Morrissey
Len & Lisa Valenti
Mary Williams
Mr. & Mrs. John Youells
Mary Anne Zezza

Kevin Scott

Mr. & Mrs. William Urban

Steve Sullivan

Kathy Kersh
Katherine Touchstone

IN HONOR OF:

Barbara Bernstein

Bob & Betty Berris
Betty & Seymour Simmons

William & Sandra Ettelson

Mike & Joan Wald

Shirley Garmer

Richard Cabeen

Mr. & Mrs. Joe Gelman

Leon Gelman

Rosemary Mills

Herbert Rorke

Ophir Trigalo

Dick & Joanne Hoffman
Roselle Shafer

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: _____



Ask the Doctor

Dear Doctor:

My neurologist said he was at a recent conference and learned that intravenous prednisone had been very helpful in treating the fatigue associated with HNPP. I started this today. I will take 250 ml once a day for 6 days, then once a month after that if it helps me. It has given me more energy and I am so thankful for that. I actually was able to walk around the block with my husband and go to dinner with my sister. Yesterday, I could hardly walk 50 feet without being exhausted.

Dr. Michael Shy responds:

The tricky thing about steroids is to separate out the energy boost which people get when they first start taking them from a true therapeutic effect. Similar boosts often occur with intravenous immunoglobulin. Usually these treatments are effective in patients with immunologically induced diseases—not genetic diseases. I am not aware of a study of pulse steroids in HNPP. It's worth pointing out that daily steroid use is associated with weight gain, diabetes, hypertension and other side effects, so people should be careful with these medications.

Dr. Gareth Parry writes:

One of the most common side effects of high-dose steroids is euphoria. I commonly use pulsed high-dose steroids for autoimmune neuropathies and sometimes for multiple sclerosis and the patients often note euphoria. Some complain of it, but most say they wish they could feel like that all the time. Paradoxically, a small number of patients become severely depressed and a few become overtly psychotic, so the euphoria is not invariable and there may be very serious consequences. In addition, even monthly doses of steroids can result in osteoporosis and osteonecrosis, although the skin changes, weight gain, fluid retention, diabetes, hypertension, etc. are less of a problem with these pulsed regimens. The bottom line is that I don't believe that the treatment is entirely safe and because it has no influence on the underlying disease, I would neither use nor recommend it for patients with HNPP.

Dear Doctor:

I have recently been diagnosed with CMT. It is most likely axonal but has not yet been genetically tested. I'm 65 and was extremely fit and agile through 1995, but since then have had gradual loss of foot strength. I still walk O.K., but with short, clipped "drop foot" style with both legs and very gradually increasing difficulty. Looking back, tiny foot surprises

such as an occasionally right numbish big toe and weakish ankle probably signaled the beginnings of disease back in 1986.

Two months ago I started taking HCTZ/25 mg and just added Zestril/10 mg which seems to have taken me out of the moderately high blood pressure category (150/105) to the O.K. range (120/80).

Through 1983 my blood pressure through good and bad times alike was always normal (120/80 maximum). Might the increase in blood pressure be disease-related—fighting cold feet, numbness and muscle degeneration? Am I better off with less medication and a slightly higher blood pressure?

The benefits of reducing high blood pressure are well established, and people with CMT are not different from other people in this regard. Thus, 150/105 is too high and should be treated as aggressively as needed, until a more reasonable blood pressure is achieved; one typically aims for 140/95 or lower. CMT is unlikely to contribute to the higher blood pressure.

Dear Doctor:

I'm wondering if anyone has ever said they had numbing of one side of the face, with pins and needles or tingling with it that lasts for about 5 to 10 minutes then goes away—but comes back again, too?

The distribution of symptoms sounds like the trigeminal nerve, but it is difficult to understand the intermittent nature of the symptoms. The patient should see a neurologist.

Dear Doctor:

What sort of promise does stem cell research hold for people with CMT? Is this our best shot for a "cure"?

Is it likely that CMT will be put "on the back burner" as far as stem cell research (because it is rare and, therefore, wouldn't generate much revenue for the drug companies)?

Finally, will the CMTA be funding any stem cell research?

The whole question of stem cell research was discussed in great detail at a symposium at the American Neurological Association in Chicago and it is, indeed, possible that genetic neuropathies such as CMT may be helped. However, we are a long way off from human application of animal studies and such studies are likely to be very expensive and need government support. I can certainly conceive of our supporting post-doc fellows working in this area and even whole projects if we had large enough donations.



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

is published by the Charcot-Marie-Tooth Association, a registered non-profit 501(C)(3) health organization. Copyright 2001, The CMTA. All rights reserved under International and Pan American Copyright conventions. No part of this newsletter may be reproduced in any form or by any electronic or mechanical means, including information storage and retrieval systems, without permission in writing from the publisher.

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

Non-Profit Org.
U.S. Postage Paid
Glen Mills, PA
Permit No. 10