

# The CMTA Report

DECEMBER 2002

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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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## Advances in the Diagnosis of Charcot-Marie Tooth Disease

**C**harcot-Marie-Tooth (CMT) is the most common inherited neuropathy and affects approximately 150,000 individuals in the United States. Affected individuals may experience muscle weakness in the arms or legs, deterioration of muscle associated with mild-to-moderate sensory loss, high-arch feet, flat feet, and depressed tendon reflexes.

**CMT genes involve 3 types of inheritance: dominant, recessive, and x-linked.**

The genes known to be associated with CMT involve three modes of inheritance: dominant, recessive, and X-linked. In order to understand these inheritance patterns, a brief overview of chromosomes is necessary. The human body is made up of millions and millions of cells. Each cell contains 46 chromosomes in 23 pairs. The first 22 pairs of chromosomes contain the same information in both

males and females. The last pair of chromosomes, the X and Y chromosomes, contains the genetic information that determines whether a person is male or female. A male has one X chromosome and one Y chromosome and a

*(continued on page 4)*

### CATEGORIES OF CHARCOT-MARIE-TOOTH

CMT is categorized by type, mode of inheritance, clinical presentation, and gene involved. The various types of CMT are provided in the table below.

**Table 1: TYPES OF CMT**

Type	Severity	Nerve conduction velocities	Gene
<b>CMT1</b>	Mild to severe	Slow (<38 m/s)	PMP22, MPZ, EGR2
<b>CMT2</b>	Mostly mild	Normal or near normal, occasionally mildly slow (>38 m/s)	KIF1B, CMT2B, CMT2C, CMT2D, NF-L
<b>Déjerine-Sottas Neuropathy</b>	Severe, begins in childhood	Very slow (<6 m/s)	PMP22, MPZ, EGR2, PRX
<b>CMT4</b>	Severe	Slow (15-30 m/s)	GDAP1, MTMR2, NDRG1, EGR2, PRX
<b>CMT1X</b>	Variable	Faster than CMT1 NCVs, but some overlap between the two types	Cx32
<b>HNPP</b>	Mild	Mildly slowed, conduction blocks	PMP22

**Abbreviations:** PMP – Peripheral Myelin Protein; MPZ – Myelin Protein Zero; EGR – Early Growth Response; KIF – Kinesin Family Member; NF-L – Neurofilament Light Chain; PRX – Periaxin; GDAP – Ganglioside-Induced Differentiation-Associated Protein; MTMR – Myotubularin-Related Protein; NDRG – NMYC Downstream-Regulated Gene; Cx – Connexin

FISCAL YEAR, JULY 1<sup>ST</sup>, 2001 TO JUNE 30<sup>TH</sup>, 2002

# President's Report

By ANN LEE BEYER, Chairman and President CMTA

## *The CMTA... Living our Mission*

**Mission Statement:**  
To generate the resources to find a cure, to create awareness, and to improve the quality of life for those affected by Charcot-Marie-Tooth.

**Vision Statement:**  
A world without CMT.

*Left to right:  
Dr. Robert Lovelace,  
Chairman/President  
Ann Lee Beyer,  
Dr. Peter DeJonghe,  
and Executive Direc-  
tor Charles Hagins  
staffed the booth at  
the International  
Congress on Neuro-  
muscular Diseases.*

This has been an excellent year for the CMTA. Our membership has grown by 11%—a significant increase. Financially, we are quite healthy, thanks to our membership increase, but also because of donor pledges, bequests, support for Charcot-Marie-Tooth (CMT) research, and special fundraising events—especially our golf tournaments—and this year, a very successful swimming event.

In addition, mainly because of the efforts of the CMTA, major breakthroughs are continuing to take place in CMT research.

This year, in order to build a more effective organization, the CMTA Board of Directors decided to combine the offices of President and Chairman of the Board, and Secretary and Treasurer. In addition, the board eliminated the office of Vice-President and added an office of Vice-Chairman. The Executive Committee consists of the President/Chairman, the Vice Chairman, and the Secretary/Treasurer. We have also added two new board members. The CMTA board is small, but we operate as a cohesive unit and we are functioning extremely well.

Needless to say, our committees are functioning well, too, especially our Executive Committee, but also the By-laws Committee, the Nominating Committee, the Research Committee, and the Fundraising Committee. We are also finding that creating task forces to carry out certain assignments is very satisfactory.

With our new structure and our very committed executive director, we are finally at a

place where we can put together a strategic plan. As part of the plan, we have looked at our entire organization, strengths and weaknesses, opportunities and threats, as well as our mission and vision, and, most importantly, where we want to be in the next few years and how we plan to achieve these goals.

The strategic planning process has been a very positive endeavor. We have many strengths, among them a very committed, active board of directors, an international medical advisory board of CMT clinicians and researchers, an excellent staff, a national support group program, a number of publications to help both the lay and medical communities understand our disease, as well as a web site, and our postdoctoral research fellow program.

The plan will be finalized and voted on at our October annual meeting. This is a major accomplishment and will give us the direction we need to grow and expand.

Unfortunately, CMT, like many orphan diseases, still remains somewhat of a mystery, not just among lay people, but also in the medical and research communities. We are actively working to change this—particularly in the medical and research communities—since they are the people we rely on for proper diagnoses, treatments, and a cure. We staff booths at various medical conferences, where we distribute information about CMT and CMT research. This year we maintained booths at the American Neurological Association, the American Academy of Neurology, and the American Society of Human Genetics meetings. In addition, we also had the opportunity to staff a booth at the 10th International Congress on Neuromuscular Diseases, held in Vancouver, Canada.

Another strength is our ability to bring leading CMT researchers and clinicians together in scientific conferences to share and discuss their knowledge and theories about this little-known disease. As many of you know, in our short existence, we have sponsored two very successful international CMT symposia. This is one of our most important strengths.

We know that we are making a difference, as we have seen the number of journal articles and breakthroughs increase significantly after



each meeting. This was brought home to us at the Vancouver meeting, when a CMT researcher from the Czech Republic told us that he had attended our Third International Conference on Charcot-Marie-Tooth Disorders and was so impressed with our organization that he went home and developed a similar organization.

Our opportunities are many: to take this orphan disease and make it a known entity, to reach more CMT patients, raise more money for CMT research, add new support groups, expand our physician education program, and establish several CMTA Centers of Excellence.

This year we have received six grant proposals for our 2003 postdoctoral fellowships. All are from researchers in leading institutions. They are in the process of being evaluated by the Grant Review Committee.

(continued on page 12)

## FROM THE EXECUTIVE DIRECTOR

As the relatively new Executive Director of the Charcot-Marie-Tooth Association, I'm constantly amazed at all the things that factor into the success of a non-profit. In a perfect world, there would always be enough income to cover the association's operational expenses. In this perfect world, memberships would always be increasing and donations would flow like honey...in a normal year and a perfect world.

But I realize that I don't live in a perfect world and that there's no such thing as a normal year. Instead, we have constant challenges, and those challenges require increasing operating revenues. For the CMTA, operating revenues come from memberships, generous contributors, and most of all, our Annual Appeal.

Everyone's world has been challenged in the past eighteen months. Between the tragedies of 9/11, the sniper shootings in Maryland and Virginia, and the sagging economy, our world is not the one we were living in only a short time ago. We know, because of this, that many of you are looking for a meaningful way to make a difference in this ever-changing and always challenging world.

When you receive this year's Annual Appeal letter, please consider our efforts to help you and give generously. Help us carry out our mission of making a difference in a person's life...that's our definition of success!

—Charles Hagins

# 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
<b>Charcot-Marie-Tooth Disorders: A Handbook for Primary Care Physicians</b>		active members \$15 inactive members \$20	
Membership Dues		\$35	
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	
<b>NEW! CMT Facts V</b>		active members \$12 inactive members \$15	
<b>A Guide About Genetics for the CMT Patient</b> <i>No shipping and handling on this item only.</i>		active members \$4 inactive members \$5	
<b>NEW! Golf Shirt</b> Size: <input type="checkbox"/> M <input type="checkbox"/> L <input type="checkbox"/> XL <input type="checkbox"/> XXL		\$15	
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	
<b>Contribution to CMT Research</b> <i>10% will be applied to administrative expenses.</i>			
<b>Shipping &amp; Handling</b> <i>Orders under \$10 add \$1.50, orders \$10 and over add \$4.50</i>			
<b>TOTAL</b>			

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 or Fax to 610-499-9267.

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.

## WAYNE STATE CLINIC NUMBER CORRECTION

The phone number for the CMT Clinic at Wayne State is 1-313-577-1689 instead of "677" as it appeared in the last issue of the newsletter.

## ADVANCES IN THE DIAGNOSIS OF CMT

(Continued from page 1)

female has two X chromosomes. Therefore, while the father can pass on either an X or Y chromosome to his child, the mother passes on only an X chromosome. If a child inherits an X chromosome from the father, the child is female. If a child inherits a Y chromosome from the father, the child is male.

Chromosomes are made up of genes that carry the information needed for making proteins. Proteins provide the structural components of cells and tissues, as well as enzymes used to initiate biochemical reactions. Genes come in pairs, in which one comes from the mother, and the other from the father. Genes are what determine everything about us, and each person has small, distinctive changes in their genes that make them unique. However, sometimes individuals have genetic alterations (called mutations) that are associated with genetic conditions. Many gene mutations have little or no effect on the health of an individual, but some mutations may affect the body's ability to properly make certain proteins and, as a result, the individual is affected with a genetic condition.

### DOMINANT

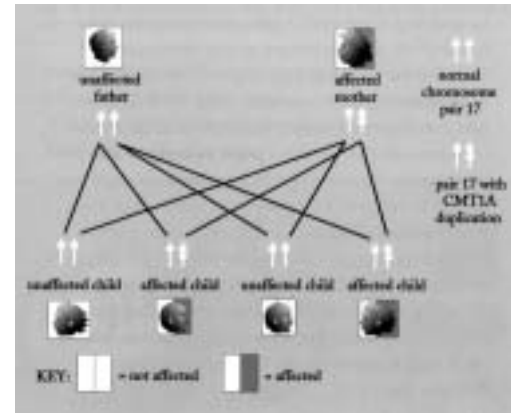
If CMT is inherited in a dominant (see Figure 1) pattern, then inheriting one mutated gene from one parent causes an individual to have or develop the condition. A parent with a dominantly inherited condition has a 50% chance of passing the mutation on to each of his/her children. Therefore, dominant conditions are usually seen in every generation.

### RECESSIVE

If the condition is recessive (see Figure 2), a person affected by the condition inherited a copy of the mutated gene from each parent. In most cases, each parent has only one copy of the mutated gene and is called a carrier. A carrier of a condition is healthy and is not predisposed to developing CMT. However, a child of two carriers has a 25% chance of inheriting the condition. Since recessive disorders are caused by the inheritance of an altered gene from both parents, not every generation is affected and it can appear that there is no family history of the condition.

Figure 1.

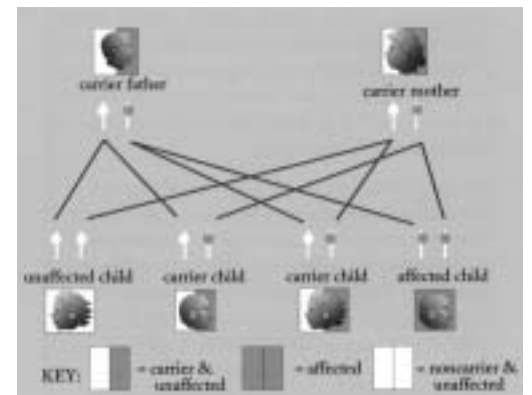
### AUTOSOMAL DOMINANT INHERITANCE



*In autosomal dominant inheritance, one parent (in this case, the father) carries a mutated gene that causes CMT. A child of these parents has a 50% risk of CMT.*

Figure 2

### AUTOSOMAL RECESSIVE INHERITANCE



*In recessive inheritance, each parent carries a mutated gene that causes the condition but is unaffected by the disorder. A child of these parents has a 25% risk of developing the CMT.*

**Table 2: SUMMARY OF CMT TYPES, INHERITANCE, GENES AND TEST AVAILABILITY**

Type	Genotype	Chromosome Locus	Inheritance Pattern	Gene	Test Availability
<b>CMT1</b>	A	17p12	Autosomal dominant	<i>PMP22</i> duplication	Yes
	B	1q21-q23.3	Autosomal dominant	<i>MPZ</i>	Yes
	C	Not 17 not 1	Autosomal dominant		No
	D	10q21.1-q22.1	Both autosomal dominant and recessive	<i>EGR2</i>	Yes
<b>CMT2</b>	A	1p36p35	Autosomal dominant	<i>KIF1B</i>	No
	B	3q13q22	Autosomal dominant	<i>CMT2B</i>	No
	C		Autosomal dominant	<i>CMT2C</i>	No
	D	7p14	Autosomal dominant	<i>CMT2D</i>	No
	E	8p21	Autosomal dominant	<i>NF-L</i>	Yes
<b>CMT4</b>	A	8q13q21.1	Autosomal recessive	<i>GDAP1</i>	No
	B	11q23	Autosomal recessive	<i>MTMR2</i>	No
	C	5q32	Autosomal recessive		No
	D (HMSNL)	8q24.3	Autosomal recessive	<i>NDRG1</i>	No
	E	10q21.1-q22.1	Autosomal recessive	<i>EGR2</i>	Yes
	F	19q13.1-q13.2	Autosomal recessive	<i>PRX</i>	Yes
<b>CMTX</b>	1	Xq13	X-linked dominant	<i>Cx32</i>	Yes
	2		X-linked recessive		No
	3		X-linked recessive		No
<b>HNPP</b>		17p12	Autosomal dominant	<i>PMP22</i> deletion	Yes

## X-LINKED RECESSIVE

If CMT is X-linked, then males and females will have different chances of inheriting the condition. If a male inherits the mutated X chromosome from his mother, he will most likely develop or be predisposed to developing the condition. On the other hand, if a female inherits the mutated X chromosome, from either her mother or father, then she will be a carrier of the condition and may have mild or no symptoms. Females that are carriers have a 50% chance of passing the mutated gene to each offspring. Affected males pass the mutant gene to all female offspring and none of the male offspring.

## SUMMARY

The discovery of genes and their functions has led to the development of genetic tests for CMT. Genetic tests can be performed on patients' blood, and Athena Diagnostics (1-800-394-4493) offers a variety of genetic tests that may detect the cause of CMT. In order for such tests to be performed, it is important to understand the benefits and

limitations of genetic testing. The benefits of testing are confirming the disease in a borderline case, the possibility of knowing the exact cause of the disease, understanding the inheritance pattern in the family, and estimating the general progression of the disease over time. In addition, once presence of a mutated gene is confirmed by genetic testing in one person, other at-risk family members can get tested for that particular gene to evaluate their risk of developing the disease. A genetic counselor is usually a good source of information on this subject, and can help identify the at-risk persons in a family.

*For a referral to a genetic counselor in your area, contact the National Society of Genetic Counselors at 610-872-7608 or online at [www.nsgc.org](http://www.nsgc.org)*

# GIFTS WERE MADE TO THE CMTA

## IN MEMORY OF:

**Bernie Bernstein**  
Barbara & Robert Bernstein

**John Allen Darnell**  
Katherine Touchstone

**W. Earle Dingman**  
Carol & Bill Rounds

**Frank Eckman**  
Forman & Betty Applegate  
Bob & Joan Lane

**Priscilla Eldredge**  
Mr. & Mrs. Douglas Moody

**Elizabeth Esch**  
Elizabeth Esch Memorial Fund and  
Martha Schott  
Terrie Hoar

**Henry Friedmann**  
Agnes Aronsohn  
Jeff & Evelyn Aronsohn  
Joslyn M. Biggins  
Erika R. Campbell  
Co-workers Ace American Ins. Co.  
Martha Tod Dudman  
Wayne Elvidge  
Sarah Emerson  
Lou & Lois Gerber

**Henry Friedmann (cont.)**  
Lorry & Irv Gottschalk  
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Ellen Kappes  
Mr. & Mrs. Jeff Manasek  
Mrs. Alfred Marqusee  
William Musselman Family  
Karen L. Peters  
Rebecca A. Pratt  
Gerald & Judith Pronko  
Annette & Alfred Rice  
Mrs. Jack Schoenfeld  
Carl Stocking  
Heidi H. Welch

**Gladys Goldstein**  
Marvin & Adele Greenwald

**Burton Gottesfeld**  
Barbara & Robert Bernstein

**John David Hill**  
Ernest & Gwen Sullivan

**Mari Ivener**  
Lauren Katz

**Helen Kiprais**  
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**William Peirce**  
William Weil

**Edward Smith**  
Maureen Kupniewski

**Ron Souser**  
Doris & Ralph Hellman

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Carol West

**Robert Williams**  
John Casazza

**Wanda Williamson**  
Kathy Carr & Katherine Touchstone

## IN HONOR OF:

**Lisa Curtis**  
A Friend

**Patricia Davis**  
Richard & Margaret Davis

**Renee Gelman**  
Leon Gelman

**Lavon Philips**  
Verne Philips

**Catherine Salerno**  
Dr. & Mrs. James Trezza & Family

### THANK YOU!

Congratulations to Chris Scarduzio and his family on the success of their golf tournament in memory of his father, J.J. Scarduzio. In their second year, they netted \$23,658 dedicated to research on CMT.

## 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: \_\_\_\_\_

\_\_\_\_\_

# Nerve Conduction Velocity Test

**N**erve conduction velocity testing (NCV) is used to evaluate damage or disease in peripheral nerves. In this test, electrical impulses are sent down the nerves of the arms and legs. The electrical impulse is applied to one end of a nerve. The time it takes to travel to the other end of the nerve is measured.

A nerve conduction velocity test is usually ordered to diagnose or evaluate nerve injury in a person who has weakness in an extremity, nerve injury, or disease (as opposed to muscular injury or disease) the severity of nerve injury, or the response of a nerve disease to treatment.

NCV testing is done by a neurologist or physiatrist. It can be done in a nerve study clinic office or at the hospital bedside, and usually takes 15 to 30 minutes.

A conducting paste is placed on patches called electrodes. A recording electrode is placed on the skin over the particular nerve in question. This electrode will record the activity or reaction of the nerve. Other electrodes are placed in a particular order near the first electrode. A special instrument is used to stimulate the nerves being studied by delivering a small shock. The recording electrode records the time it takes for the shock to cause activity in the nerve.

The time is sent to a machine called an oscilloscope. This machine can measure the response time of the nerve to stimulation. A calculation is then done on the response time and the distance between the electrodes. The electrical stimulation may be slightly uncomfortable during the test.

Normal results from a nerve conduction velocity test mean that there is no evidence of damage or disease in the peripheral nerve. Nerve damage or disease may still exist despite normal NCV results. This is because other healthy fibers in the same nerve may show a normal reaction time. The test is often done in conjunction with electromyography (EMG) to evaluate neuromuscular abnormalities.

Abnormal results may depend on why the NCV test is being performed. These results may indicate:

- peripheral nerve injury or disease
- carpal tunnel syndrome
- poliomyelitis, a condition that affects the spinal cord
- diabetic neuropathy, temporary or permanent nerve damage due to diabetes

- herniated disc
- myasthenia gravis, a disease that causes extreme muscle weakness and fatigue
- Guillain-Barré syndrome, a condition that causes nerve damage and inflammation, and muscle weakness and paralysis
- damage to a nerve from trauma
- chronic inflammatory polyneuropathy, the simultaneous malfunction of many peripheral nerves throughout the body.

## Essays Wanted

**G**uidelines For a Different Journey: Personal Stories for Parents by Adults with Disabilities is a new book that Stan Klein and John Kemp are co-editing. For this book, adults who have grown up with disabilities and/or health care needs are invited to write short essays for parents of children with disabilities and/or health care needs. In their essays, authors are asked to write an essay that they wish their own parents had shared with them while they were growing up.



Here are specific guidelines for essays:

1. Please write an essay of about 1500 words or less. Add a biography of about 150 words or less that would follow the essay in the book. At the end of the essay, please write your mailing address, telephone number, fax number, and e-mail.
2. Please submit your essay as an attachment in Microsoft Word to an e-mail or paste your essay into the body of your e-mail. Send e-mail to: stan@disabilitiesbooks.com
3. If you submit your essay by regular mail, please double space the text. If at all possible, please also submit the essay on a disk as well. Please label each page of your printed essay and the disk with your name and address. Please send the printed copy and disk to: Stanley D. Klein, Ph.D., DisABILITIESBOOKS, Inc., P. O. Box 470715, Brookline, MA 02447-0715.
4. Deadline: All essays are to be received by February 15, 2003.

*Authors of essays accepted for inclusion in the book will receive \$125 for the right to include their essay.*

# The Role of Physiotherapy in the

By **PAUL VAN DER DOLDER**, Senior Physiotherapist, Concord Hospital, Sydney, Australia

*This article is an update of an article by Julianne Wheatley, a physiotherapist who has worked extensively with CMT patients, and is reprinted here with permission of the Charcot-Marie-Tooth Association of Australia. Please remember that this is the advice given in Australia. You should check with your own physician before beginning any new program.*

**C**MT affects the sensation and muscle control of the nerves of the lower legs and lower arms. People with CMT commonly complain of difficulty walking, especially on uneven ground: catching toes and tripping (and occasionally falling), poor balance, going over on their ankles, and weakness of the hands.

The difficulties with walking and running, the tripping and falling, and ankle sprains are generally due to muscle imbalances in the lower leg. That means that some of the muscles of the leg become weaker due to CMT, while the muscles that are still working in the other direction tend to become shortened.

The muscles of the lower leg can be divided into two groups—the anterior (front) and the

**If some of the muscles are weak in the lower leg, then you will usually have impaired balance.**

posterior (back) muscles. In CMT, the anterior muscles are primarily affected first. The main muscle in the front of the leg is the tibialis anterior muscle, which pulls the foot up at the ankle. When this is affected the patient has trouble picking up the foot, which leads to a 'high stepping' style of walking, so that the foot clears the ground and the weakness may lead to increased tripping. Some other muscles are also affected by the same muscle. These muscles, peroneus longus and brevis, run on the outside border of the lower leg and turn the foot up and out. They also prevent the foot from turning in ('going over on the ankles').

The posterior compartment (the calf muscle) is made up from the plantar flexors (those muscles that pull the foot down) and the invertors (those that turn the foot in). These muscles are affected at a later stage.

As a result of the weakness of the muscles on the front and outside of the leg, the person with CMT may have difficulty lifting his foot up to clear obstacles such as stairs or the edge of rugs. He may also tend to turn the foot in more as the muscles on the outside of the leg are not functioning properly.

Muscle imbalance may also lead to tightness of the opposing muscles. In patients with CMT this is usually the calf muscles and the muscles that turn the foot in (the invertors). As these muscles get tight, the patient may become aware that it is more difficult to stand with the heels on the ground. This also makes lifting the foot much more difficult. Tightening of some of the muscles within the foot and toes results in deformities of the feet, e.g., claw toes and high arches. This can cause pain and difficulty when walking.

If some of the muscles are weak in the lower leg, the patient will usually have impaired balance. This is often due to the lack of the ability in the muscles around the ankle to support

**Figure 1**  
**CALF STRETCH ON WEDGE**



Stand on a wedge with your back against the wall. Keep your feet firmly on the wedge, knees straight and shoulders against the wall.

**Hold this position for 20-30 minutes.**  
**Do one session a day.**



# Management of CMT

it. Balance sensation is felt by sensory receptors in the joints and these are affected as the nerves to the feet are affected. The patient's ability to feel what position the ankle is in is, therefore, affected which, also affects balance.

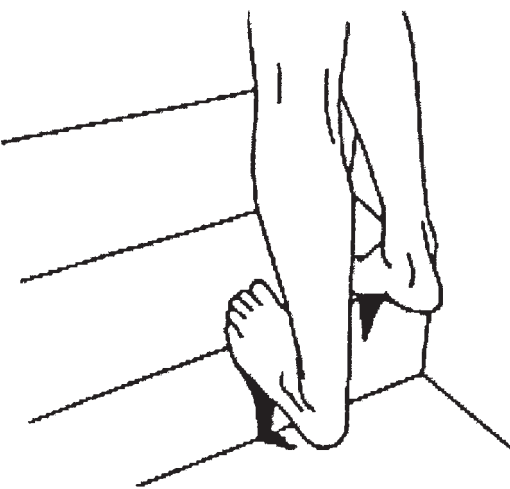
The goals of exercises with patients with CMT are, therefore, to:

1. Maintain or improve muscle strength—in order to:
  - a. maintain the function of the muscles so that everyday tasks such as walking can be continued
  - b. minimize injuries by improving stability
2. To maintain or improve balance
3. To stretch out muscles that may have become tight
4. Maintain or improve cardiovascular conditioning.

There is no standard recipe for exercises with CMT. Some of the commonly prescribed exercises given to CMT patients are illustrated in Figures 1–5.

**Figure 2**

## ARCH STRETCH ON STEP

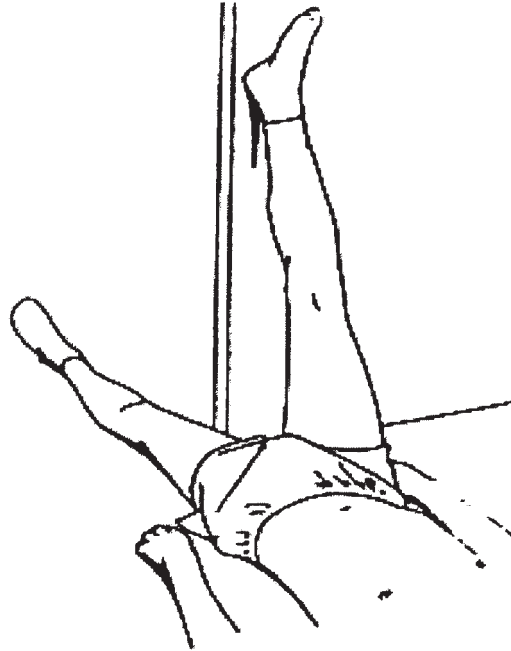


Stand with the ball of the foot on stair, reach for bottom step with heel until a stretch is felt through the arch of the foot.

**Hold for one minute. Relax. Repeat 10 times.**  
**Do one session a day.**

**Figure 3**

## HAMSTRING STRETCHES



Lying on the floor with the involved leg on the wall and the other leg through a doorway, scoot buttocks toward the wall until a stretch is felt in the back of the thigh. As the leg relaxes, scoot closer to the wall.

**Hold this position for 10 minutes.**  
**Do one session a day.**

## STRETCHING EXERCISES

These help to prevent tightening of muscles and to help lengthen already tight muscles. All stretches are specific for each muscle, so general stretching exercises are not sufficient. To be effective, muscles need to be stretched for at least 10 to 15 minutes with a sufficient amount of force applied. Commonly prescribed stretches include calf stretch on wedge (figure 1), arch stretch on step (figure 2), and hamstring stretches (figure 3).

If possible, try and do something that you enjoy while doing these stretches, e.g., read a book, watch TV, listen to some music. This will

*(continued on page 10)*

## PHYSIOTHERAPY AND CMT

(Continued from page 9)

help the time pass. It is best if these stretches are done on most days of the week.

Stretches are probably the most important part of an exercise program and will help to prevent foot deformities and calf tightness, which will, therefore, help maintain a pain-free, functional gait.

## STRENGTHENING EXERCISES

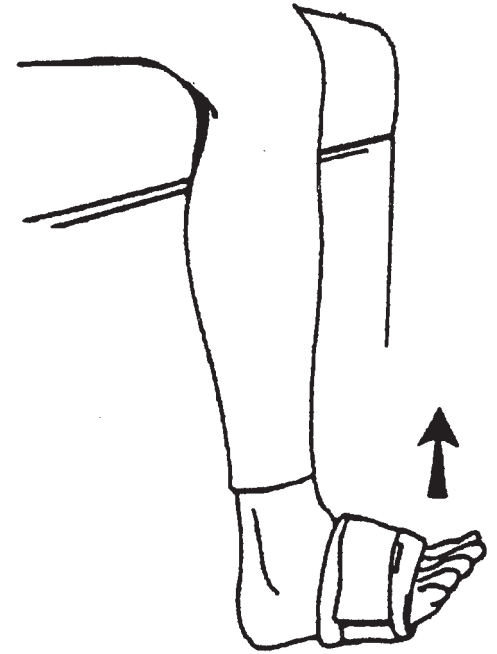
These are best done below your maximum capacity (i.e., don't exhaust your muscles). Strengthening exercises are usually most effective when commenced early in the disease, when the muscle fiber degeneration and weakness are less. Some common strengthening exercises that we prescribe are dorsiflexion strengthening with a weight (figure 4) and eversion strengthening with a weight (figure 5).

## BALANCE ACTIVITIES

Any activities that challenge balance are good for retraining. Some examples are throwing and catching a ball, standing on one leg, and standing on a “wobble board.”

Picture No. 4

## DORSIFLEXION STRENGTHENING WITH WEIGHT

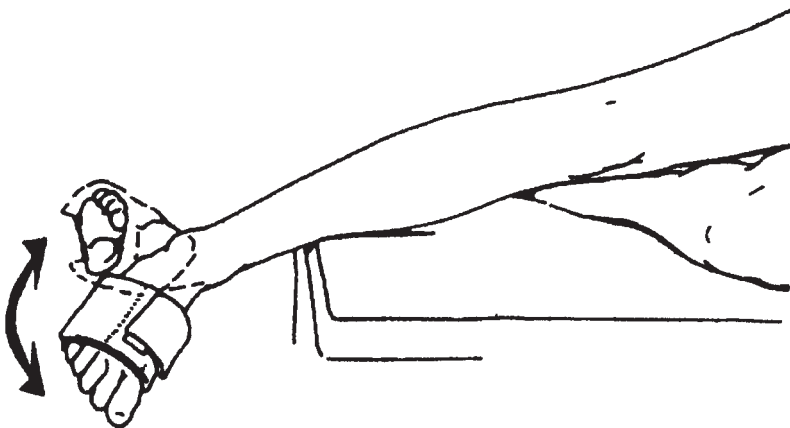


Sitting with leg over the edge of a table or bed with a bag or rice wrapped around the foot, bend the ankle/foot up toward the knee.

**Repeat 10 times per set**  
**Do two sets per session**  
**Do two sessions a day**

Figure 5

## EVERSION STRENGTHENING WITH WEIGHT



Lying on one side with a bag of rice wrapped around the foot, bend the ankle/foot up and turn the foot out.

**Repeat 10 times**  
**Do two sets per session**  
**Do two sessions a day**

## GROSS MOTOR ACTIVITIES

Young children with CMT tend to display difficulty with some basic gross motor skills, e.g., running, hopping, and climbing. With practice, these skills can improve and should be encouraged.

## SPLINTS

These take on two forms—assistive and corrective. Assistive splints include AFOs (foot-drop splints), which are worn inside shoes and act to hold the front of the foot up to prevent tripping. Corrective splints are night splints that hold the foot up at night to provide a mild stretch over a long period. These may be useful for some children with CMT.

All of these treatments, although prescribed by a physiotherapist, are your responsibility. For them to be effective you must do them regularly—once every couple of days is not enough. Try to build your exercises into your daily life—do your stretches while watching television, reading a book or listening to music.

# Vocational Rehabilitation... Useful for CMT Patients

**V**ocational rehabilitation (VR) was developed during the late 1800's when various government agencies tried to help disabled veterans find jobs. In 1920, Congress passed the Civilian Vocational Rehabilitation Act.

Vocational rehabilitation is a program designed to help disabled people become fit for employment. Vocational rehabilitation programs are generally designed for people aged 16 and older who have physical or mental disabilities. There are three activities in vocational rehabilitation: 1) rehabilitation counseling, 2) vocational evaluation, and 3) job placement.

Under the Rehabilitation Act of 1973, each state receives a federal grant to operate a comprehensive VR program. The state-operated program is designed to assess, plan, develop, and provide VR services to eligible individuals with disabilities, consistent with their strengths, resources, priorities, concerns, abilities, capabilities, interests, and informed choice. By providing services in this way, the VR program enables individuals with disabilities to prepare for and engage in gainful employment.

To be eligible for VR services, a person must be an individual with a disability, which means that he or she has a physical or mental impairment that constitutes or results in a substantial impediment to employment, and he or she can benefit from VR services and achieve an employment outcome. Anyone who receives Supplemental Security Income (SSI) or Social Security Disability Insurance (SSDI) benefits is presumed to be automatically eligible for VR

services, unless he or she is too severely disabled to benefit from the services.

The kinds of services available through Vocational Rehabilitation include the following:

- an assessment for determining eligibility and VR needs
- vocational counseling, guidance, and referral services
- physical and mental restoration services
- vocational and other training, including on-the-job training
- maintenance for additional costs incurred while the individual is receiving VR services
- transportation related to other VR services
- services to assist students with disabilities transition from school to work
- rehabilitation technology services and devices
- supported employment services
- job placement services.

On the basis of an individual's financial resources, the state VR agency may require an eligible individual to help pay for services. However, the following services are available to all eligible individuals, regardless of their financial resources, without charge:

- assessments to determine eligibility and VR needs
- vocational counseling, guidance, and referral services
- job search and placement services.

For information about the VR agency in your state, check the local phone directory. The office's phone number and address are usually listed under "State Government."

## *Good News for Members of the CMT Community*

**T**he Institute of Medicine, an arm of the National Academies, announced the election of 65 new members, raising the total active membership to 1,358. New members are chosen for their major contributions to health and medicine or to related fields, such as social and behavioral sciences, law, and economics.

A member of the CMTA's Medical Advisory Board, Dr. James Lupski, professor of molecular and human genetics, Baylor College of Medicine, was among those newly elected.

A second announcement of good news was the awarding of a contract to the Wayne State University School of Medicine to conduct studies into maternal and infant health and disease. This award from the National Institutes of Health will bring more than \$100 million to the School of Medicine and the city of Detroit over the ten-year life of the contract. This announcement is an honor to the School of Medicine and will increase the resources and standing of the institution.

## Reaction and Response

In the latest issue of The CMTA Report, I read with great interest the personal article by J. D. Griffith about the untimely death of his teenage daughter, Marah. But it was the poem by Marah herself that jumped off the page to me. “Come Take a Walk in My Shoes” could have been the story of my life.

You see, I have walked in Marah’s shoes for over fifty years. The poem brought back devastating memories and with them, tears. I read the poem over and over again. I, too, would walk down the street and have people stare at me and my gait. It hurt me both emotionally and physically. The pain was doubled.

Living through the teenage years is bad enough, but having CMT, which can cause visible signs of difference, was horrible. All I wanted was to be like everyone else. I wanted peer acceptance; I wanted to date; I wanted to compete in athletics. But, I wasn’t like all my friends and it was a very difficult situation to accept.

So, now years later, I still feel as though I walk in Marah’s shoes. It doesn’t really get any

easier to accept the pain, the gawking, or the limited mobility; however, the passage of time and the perspective of “age” coupled with the support of my family and friends and some excellent medical care have helped me. I’ve become an active member of the CMTA’s Board of Directors and an advocate who works to educate the public about our little-known disorder.

I’m working hard to make a difference for CMT patients today and for future generations, among them my own children and grandchildren. Yes, I still walk in Marah’s shoes, as I know many of you do, as well. But we can make a difference. Let’s not just walk in Marah’s shoes, but let’s work to change things by contributing in Marah’s memory to the CMTA’s research fund so that future Marahs might not need to write such painful and touching poems about their feelings of being different.

—Richard Sharpe

## PRESIDENT’S REPORT

*(Continued from page 3)*

Our CMT North American Database project generated a great deal of interest at the Vancouver neuromuscular meeting. At the end of its first year, over 500 forms have been received and 234 people are now registered. We are ahead of schedule. The purpose of the database is to gain a better understanding of CMT so that CMT researchers can have access to large amounts of information about CMT patients. We are happy to report that information from the database is now available for qualified CMT researchers.

Another major accomplishment of the CMTA in 2001–2002 is the establishment of a CMT North American Consortium. This meeting is open to CMT investigators with expertise in molecular biology, genetics, and neurophysiology. Its purpose is to foster communication and collaboration among clinicians and researchers.

Our Medical Advisory Board meets several times a year, usually at the neurology and genetics meetings. Headed by Dr. Robert Lovelace, Neurological Institute, Columbia Presbyterian, this board, too, is quite active.

Under Dr. Lovelace’s direction, it has established several task forces. The job of the first task force, headed by Dr. Gareth Parry, University of Minnesota, is to develop a list of criteria for establishing CMTA Centers of Excellence, which will be presented at our consortium meeting this winter. The second task force, Chaired By Dr. Jack Petajan, University of Utah, and Dr. Lisa Baumbach-Reardon, University of Miami, has completed its review of the Physicians Handbook for Primary Physicians, and has recommended the addition of several new chapters. Dr. Michael Shy, Wayne State University, and Dr. Angelika Haan, London (Ontario) Health Science Center, are heading up a task force that has put together the program and made arrangements for the North American CMT Consortium. In addition, Dr. Florian Thomas, St. Louis University, Chairman of the Grant Review Committee, has evaluated the grant review policy and had revised the evaluation process.

We are very excited about the future and the possibilities it presents and are deeply committed to living our mission: “To generate the resources to find a cure, to create awareness, and to improve the quality of life for those affected by Charcot-Marie-Tooth.”

# The Herb of the Month: *Kava Kava*

By **BRUCE A. CRISTOL, PharmD**

**N**ative to the islands of the South Pacific, Kava Kava has been used for centuries as a beverage to induce relaxation. In fact, scientists have detected traces of Kava Kava on ancient cultural items unearthed by archeologists. The famous British mariner and explorer Captain Cook named the plant “intoxicating pepper” (*Piper methysticum*) and introduced it to the Western world.

Botanists describe Kava Kava as the dried horizontal stems (rhizomes) and roots of *Piper methysticum*, a large shrub cultivated in places such as Fiji. Of the twenty varieties of kava plants identified, the black and white grades are the most highly prized.

Hawaiians have used Kava Kava for asthma. However, the most prominent use of the plant has been for sleep and relaxation, including

relief from anxiety. Other uses have included treatment of the common cold and other respiratory infections. Also, Kava Kava has been used orally for urinary tract infections. However, problematic adverse reactions, including

liver toxicity (see below), scaly rashes, blood in the urine, and temporary yellow discoloration of the skin, hair, and nails, have been reported.

It is important to re-emphasize the following: Kava Kava is not considered a “drug” by the U.S. Food and Drug and

Administration (FDA). Therefore, the buyer should be aware that commercial preparations, such as extracts, and topical preparations may vary from dose to dose, batch to batch, or manufacturer to manufacturer.

Drug-herb interactions reported in the medical literature are shown below.

## WARNING!

There have been many reports in the medical literature, especially from Europe, indicating that Kava Kava has been associated with liver damage and even death. Some patients have required liver transplants. In fact, this herbal substance has been banned in Germany and Switzerland.

Drug interactions with Kava Kava	Result of interaction	Comments
Alcohol	Drowsiness Motor reflex depression Increased risk of developing liver toxicity	
Diazepam (Valium®) and others like it, e.g. alprazolam (Xanax®)	Motor reflex depression Drowsiness	
Levodopa (Larodopa®)		Reduced effectiveness of levodopa

### Use of the following drugs is not recommended due to the increased possibility of liver toxicity:

carbose (Precose®); amiodarone (Cordarone®); atorvastatin (Lipitor®); azathioprine (Imuran®); carbamazepine (Tegretol®); diclofenac (Voltaren®); fluvastatin (Lescol®); gemfibrozil (Lopid®); isoniazid (INH); itraconazole (Sporanox®); ketoconazole (Nizoral®); leflunomide (Arava®); lovastatin (Mevacor®); methotrexate; nevirapine (Viramune®); niacin; nitrofurantoin (Macro-dantin®); pioglitazone (Actos®); pravastatin (Pravachol®); pyrazinamide; rifampin; ritonavir (Norvir®); rosiglitazone (Avandia®); simvastatin (Zocor®); tacrine (Cognex®); tamoxifen; terbinafine (Lamisil®); valproic acid (Depakene®).

There may be other medications that, when used together with Kava Kava, may have adverse effects on the liver.

### REFERENCES:

- DerMarderosian A. The Review of Natural Products. *Facts and Comparisons* 1999.  
 Jellin JM, Gregory PJ, Batz F, Hitchens, K, et al. Natural Medicines Comprehensive Database 2002.  
 Miller L. Herbal Medicinals: Selected Clinical Considerations Focusing on Known or Potential Drug-Herb Interactions. *Archives of Internal Medicine* 1998; 158: 2200-2211.

# Designing an Energy Loading

By MITCHELL S. WARNER, CPO

Currently there are many different orthoses that are being applied to the patient with Charcot-Marie-Tooth disease. They are made from a variety of materials that include metal, thermoplastics, and carbon-graphite composites. These orthoses are either off-the-shelf (pre-made) or custom made.

While thinking about what type of AFO is appropriate for the patient with CMT, we have to take into consideration some basic principles:

- 1) Static alignment
- 2) Balancing: static and dynamic mode
- 3) Increased function

The goal is to incorporate these basic principles into a custom orthotic system designed to reduce fatigue, prevent further deformity, and walk with a more stable gait.

The terms that I use in describing my designs are energy-loading and dynamic release. As more energy is loaded into the orthotic structure, more energy is dynamically released. Proper alignment and balancing will load more energy into the brace. What materials can most effectively store and return energy? With metal we are limited to spring-activated joints. These joints have very small springs that are approximately 2.6-cm in length, or about 1 inch. Their compression and energy release are small. Next we have thermoplastics. While thermoplastics are lighter in design than metal, they are not as strong. Although we can get spring, we do not have full control, because the thermoplastics torque or twist when a load is applied to them. This can allow deformity to progress.

Therefore, the best strategy is to get as much spring with maximum control. This is why my designs use carbon-graphite construction. With carbon-graphite we get a long lever to spring (approximately 2/3 the total length of the leg below the knee), in combination with rigidity to control the limb and its joints.

## CONTROL OF THE LIMB CREATES STABILITY

1) *Static control.* This means to control any foot deformity present. If a foot goes into a varus (supinated) position, present in most CMT type I patients, we want to realign the limb and correct as much of that rotational deformity as possible. We want to see this statically while bracing, as this is a structural correction.

2) *Balancing control.* This is a very interesting area with CMT because with conventional bracing we look at balance strictly in gait. We normally look for imbalance while the patient is walking. But with CMT, standing (static) balance becomes critical. Most CMT patients tell me that when they stand, they need to move their feet up and down, side to side, or front to back to prevent them from falling or grabbing something to maintain their stability. Therefore, balance with CMT becomes critical in the static mode. With carbon-graphite we can restore the center of gravity and alignment to achieve a stable standing base. This helps restore balance from side to side and/or from front to back.

3) *Functional control.* This is an outcome of the first two controls combined. Once we have a correction of the structure added to the restoration of balance, everything is put in place for the CMT patient to become more functional. If we can get a better balance when standing, this usually transfers to better balance in walking. These components should not be separated. The only exception to this rule is a rigid contracture that cannot be corrected through bracing and that might be a surgical consideration.

During the stance phase of the gait cycle, we are concerned about how the foot strikes the floor. This is commonly referred to as heel strike or initial contact.

As the orthosis is loaded with the body's weight, the goal is to have stability that allows the limb to advance in a stable line of progression.



# Brace for CMT

## THREE PLANES OF MOTION

The Helios and Helios II orthoses are designed to provide correction in all three planes of motion of the foot. This is also referred to as “triplanar control.”

Although one joint may be out of alignment, it will affect other joints, as they will make compensatory adjustments that can create further deformity. This is why it is critical to correct as much alignment as possible. Other joints that are involved in orthotic alignment are the transverse tarsal joint (midtarsal joint) and the talocrural joint (ankle joint). The transverse tarsal joint inverts and everts the foot. The talocrural joint dorsiflexes and plantarflexes the foot.

## DIAGNOSTIC FITTINGS

After I take a custom mold of a patient's limb for an energy-loading carbon graphite device, I do not then go to the fabrication of that device.

What takes place next is the modification and fabrication of a set of diagnostic (or prototype) orthoses. The diagnostic orthosis serves as a fitting tool before the final carbon-graphite device is made. A great deal of information is learned from the diagnostic because the patient stands up in it and walks in it. This gives great feedback as to the alignment, stability, and comfort of the patient. It is at this time that we check to see if we have appropriate triplanar control before the final device is fabricated.

If proper diagnostic fittings are completed and the limb is placed in a corrective alignment, most patients can achieve a more balanced and functional gait. I do not want to make anyone think that it is as simple as having the brace made, putting it on, and walking out. Like any other device that requires balance, whether it is a device for playing sports or simply walking, there is always training involved. The amount of training with the average patient is learned quickly because once the alignment and balance are restored, the patient feels much more stable, and will acclimate to the new balance. This is usually the case, as improved balance provides stability and security, with energy return.

After the diagnostic fittings are complete, new molds are made from the diagnostics. Any new alignment changes or modifications are incorporated into the new molds, and fabrication of the final carbon-graphite device begins.

## DIFFERENCES IN HELICAL ENERGY LOADING DESIGN

The Helios orthosis was designed primarily to load energy in its helical uprights and correct deformity at the same time. It corrects in all three planes of the foot. Carbon-graphite gives the orthosis rigidity for control and also the energy-loading structure to store and release energy. Helios stands for: Helical Energy-Loading Integrated Orthotic System. I currently design and fabricate two versions of the orthosis: the Helios and the Helios II. Structurally they are very similar in the way they load and release energy to the wearer. While the Helios I allows adaptation for uneven ground, the Helios II does not. Through fitting many patients with helical design braces, I have found that some like the feeling of adaptation to uneven ground, but others have commented that they would like more lateral stabilization. Therefore the Helios II was designed to provide more stability to oppose lateral movement.

The Helios II design also has adjustability for compression of the uprights, which determines how much energy is loaded and released. In both designs I quite frequently use a customized system of silicone padding to maintain correction and control in the foot. On the Helios page of our website there is a link to video clips where you can view a before and after demonstration of a CMT patient wearing the Helios orthosis. There is also a section called the “CMT-Guide”.

I often receive e-mails asking about bracing for CMT and patients wanting to know how to get an evaluation from out of state. The easiest way to do this is to send me a videotape which I will review and pre-evaluate for any CMT patient at no charge. I provide videotaping instructions for this. If you would like to contact a CMT patient who has been fit with the Helios orthosis, please contact us at:

Ortho Rehab Designs  
601 S. Rancho Dr., Suite B-14,  
Las Vegas, NV 89106  
702-388-9909  
Website: [www.ORDesignsLV.com](http://www.ORDesignsLV.com)  
E-mail: [ORDesignsLV@cs.com](mailto:ORDesignsLV@cs.com)

Ortho Rehab Designs is an independent orthotic and prosthetic facility, and is not associated with any other company that provides energy-loading or dynamic response orthoses.

## Support Group News

### ■ California - Northern Coast Counties

The August meeting featured Meganwind Eoyang who provided exercises and suggested that CMT patients find ways to do daily activities with as much playfulness and effortlessness as possible. She suggested picking up things with two hands rather than gripping hard to brace against the weight of the object.

In November, we were visited by Dr. Jerome Chinn, a well-known neurologist from Santa Rosa. Our member, Dr. Bill Finnegan was instrumental in arranging for Dr. Chinn's visit.

Group leader, Freda Brown, suffered a broken leg some weeks ago and is currently unable to host the meetings. She is recovering well and we wish her a speedy and full recovery.

### ■ Colorado - Denver Area

The last meeting was November 4, 2002, at the Glory of God Lutheran Church. The meeting was planned to discuss what direction the

group should take and what speakers, videos, and discussions we want for the coming year.

The September meeting featured Gail Feeney-Coyle, who gave an outstanding report on the visit she and her daughter made to the CMT Clinic at Wayne State University in Detroit. We are hopeful that more families will take advantage of this resource which is so helpful to those with CMT.

### ■ Oregon/Pacific NW

The October issue of the newsletter of the Pacific NW CMT support group contains articles on the differences between CMT, MS, and MD written by Dr. Greg Carter, suggestions for ways to treat callouses, hints for using smaller containers when you have weak hands, or using lever dispensers, and a lengthy article on restless leg syndrome from the Harvard Health Letter.

The group has a new website address at <http://groups.msn.com/CharcotMarieTooth>

## Johnstown Support Group Forms

The first meeting of the Johnstown, PA CMT support group was held on Saturday, September 21 with 45 people in attendance.

The meeting was held at the Crichton Rehabilitation Center, a part of the Conemaugh Health System. The meeting was an incredible experience for myself and all of the attendees. CMTA Board Chairman and President Ann Lee Beyer did a great job of answering the endless questions posed by those in attendance, and Executive Director Charlie Hagins spoke about the CMTA and the challenges of CMT.

The heartfelt support for each other was remarkable, but the unbridled enthusiasm for the cause of CMT was truly astounding. *How can I help? What can I do? Let's get moving! Call our congressman. Write our senator. Give me some more brochures to pass out. Let's get together again—soon! WOW!*

I found the response amazing as the word spread of the meeting. The CMTA office sent meeting flyers to the individuals on their mailing list and to neurologists (to post in their offices). The hospital (Conemaugh Health System) put a notice in their newsletter, sent press releases to the media, and placed a small ad in



the local newspapers and on public service channels. The most effective tool, however, was personal telemarketing. When we received a response, I called, talked about the meeting, and asked if they knew anyone else with CMT. Many knew others or had family members with CMT and offered to contact them. Others contacted their doctors and posted flyers. The biggest problem turned out to be finding a room big enough! We expected more attendees, but conflicts, problems with handicapped transportation and bad weather held attendance down.

(continued on page 19)

*J. D. Griffin, standing, presented Donald F. McLaughlin with a certificate recognizing his contributions to the cause of CMT.*





# 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—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@mac.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:** Martha Hall,  
502-696-3338  
**E-mail:** marteye@mis.net

## 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 bimonthly  
**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:** 2nd Thursday of each month  
**Contact:** Dr. David Younger,  
212-535-4314,  
Fax 212-535-6392  
**Website:** www.cmtnyc.org

## 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:** 3rd Saturday of each  
month, excluding July &  
August  
**Contacts:** Diane Kosik, 914-937-2013,  
Beverly Wurzel,  
845-783-2815  
**E-mail:** ladydismiles@aol.com or  
cranomat@frontiernet.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:** jeanie4211@attbi.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

## Pennsylvania—Johnstown Area

**Place:** Crichton Center for Advanced  
Rehabilitation  
**Meeting:** Bimonthly  
**Contact:** J. D. Griffith, 814-539-2341  
**E-mail:** jdgriffith@mail.charter.net

## Professionals Working for CMT Patients: Thomas D. Bird, MD

I am a native of the lovely Finger Lakes region of western New York. I obtained my undergraduate degree at Dartmouth College in the green hills and valleys of New Hampshire and then made a drastic switch to midtown Manhattan to complete my medical degree at Cornell. I then traveled to the beautiful Pacific Northwest and Seattle for my Internship and Neurology residency training at the University of Washington. Except for a brief tour of duty in the Navy, I have remained in the Northwest for more than 30 years.

During my Neurology training I was attracted to the field of Medical Genetics because of the large number of fascinating patients that I saw with genetic diseases of the nervous system. Dr. Arno Motulsky, a pioneer in the field of Medical Genetics, helped me see how principles of genetics could be productively applied to neurological diseases. So in the mid-1970's I became a clinical neurogeneticist before the field actually existed.

Two of the most common disorders I saw in those early days were Huntington's disease and Charcot-Marie-Tooth neuropathy. For Huntington's there was excellent data allowing us to calculate a family member's risk for having the disease based on that person's age. No such data existed for CMT. Therefore, my first CMT project was collecting and analyzing such data to use for genetic counseling of families with CMT.

Having collected a large number of families with CMT, I realized the disease was a perfect candidate for a genetic linkage study, which would allow us to identify regions on human chromosomes containing genes for CMT. Our first attempt was highly successful even though it was done with simple blood group markers, prior to the advent of the far more useful DNA markers. We assigned the first CMT genetic locus to the long arm of

chromosome 1 in 1980. This eventually became known as CMT1B. Our original family with CMT1B was subsequently used to identify the underlying gene (known as myelin P0) by Hyasaka and colleagues in Japan in 1993.

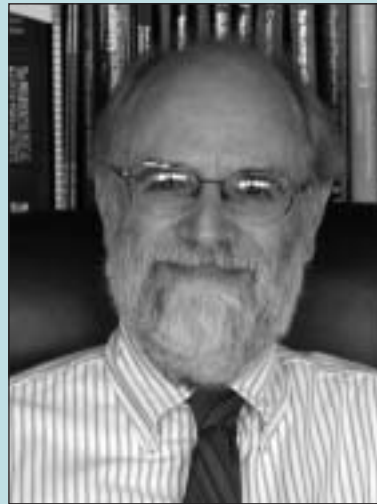
Over the years I have had the opportunity to collaborate with many wonderful people in our studies of CMT. My first collaborators were Drs. George Kraft, a physiatrist, Eloise Gibblett, a human geneticist, and Jurg Ott, a postdoctoral fellow at that time who became

a world authority on linkage analysis. Dr. Phillip Chance has been a friend and colleague for more than 20 years. More recently I have had the pleasure of collaborating with Drs. Greg Carter, Valerie Street, and Craig Bennett. Our studies have continued to be productive, including the new discovery of the gene underlying CMT1C, an entity identified by Dr. Chance and me nearly a decade ago.

My experience with CMT families has not only been a source of scientific knowledge, but has shown me outstanding examples of

marvelous human beings learning to cope with and overcome frustrating disabilities. I have visited numerous CMT families throughout the American West, often as a guest in their homes during delightful family reunions. I have had the wonderful experience of personally following three and even four generations of CMT families for more than 25 years. In fact, my own family has a member with CMT who has been an inspiration to all of us.

I can assure you that the CMT research community is full of bright, imaginative, and hard-working physicians and scientists who will continue to push back the frontiers of this fascinating and important disease. I enjoy being a part of this great enterprise.



*Dr. Thomas Bird*

# Letters to the Editor

## Dear CMTA:

I read with great interest your article, "Respiratory Issues in CMT: A Personal Story". First, I want to offer my condolences to J. D. on the loss of his daughter. I agree that the phrase, "Does not affect life expectancy" from the "What is CMT?" description in the literature should be removed.

I lost my son, Francis (Frank) Robison, at the age of 47 on 2/14/02. He had Charcot-Marie-Tooth and could not walk. He used a motor chair to get around. He never complained and "played the hand he was dealt." He developed pancreatitis brought on by a gall stone. He was back and forth between the intensive care unit and a rehabilitation hospital for 7<sup>1</sup>/<sub>2</sub> months. Every time he went to rehab he got an infection and had to be rushed back to ER and intensive care. He was on a ventilator with a trache for most of this time and, therefore, could not eat or talk. Every time they tried to remove the ventilator, he could not breathe on his own. He had surgery to remove the gall bladder and drain a pseudocyst on the pancreas. He was fed through a tube, had a drain tube in his stomach, a catheter for urine, tubes for medications, and blood transfusions. More than once when I questioned the doctors about his lack of progress, they would say, "well, he does have this neuromuscular disease." Even when he finally just started staring into space and not recognizing anyone, the neurologist was quick to blame the "neuropathy" on CMT. When he was not responding to speech therapy with the trache, they had an ear, nose, and throat doctor check the vocal chords. They were not working properly.

Thank you for letting me tell you my personal story. As you know, there is nothing more difficult than to bury your own child. Frank has a son and an 11-month-old grandson. Unfortunately, he never got to see his grandson. Keep up the good work on raising awareness and money to fight CMT.

—M. E., Maryland

## Dear CMTA:

Thank you for including the letters in the October 2002 issue of The CMTA Report about how Macrobid increases the symptoms of Charcot-Marie-Tooth disease.

Macrobid was prescribed for me by my urologist several years ago, 1<sup>1</sup>/<sub>2</sub> years before I

showed CMT symptoms and was given that diagnosis. My father had CMT; I recognized the symptoms immediately. I sent a copy of the October 2002 issue of The CMTA Report to my urologist with the first letter to the editor circled and asked him to share this information with his colleagues.

Two days ago I received a letter from my urologist, who states:

"Thank you for the information regarding the use of Macrobid in individuals who have been diagnosed with Charcot-Marie-Tooth disease. I have shared this information with my partners and appreciate your help. I hope you are doing well".

So, again, thank you for getting the information to me so I can pass it on.

—D. C.

## Dear CMTA:

Why not include a "HINTS" column of contributions from those of us who live with CMT. My contribution: If you don't want to spend \$45 on microwaveable slippers to get your feet warm, just moisten some heavy woolen socks and put them in the microwave for a minute or two. Be careful and don't burn your hands when removing them and don't microwave them so long they're ready to burst into flames.

—R. K.

## JOHNSTOWN SUPPORT GROUP

(Continued from page 16)

We were dealt our imperfect genetic hand, but we have a choice. We can feel sorry for ourselves as CMT tightens the screws on our physical abilities or we can decide to do something to change the future.

You can start a support group or join an existing one. Help yourself, your family, and a whole lot of other people by getting involved. Contact the CMTA, your local support group, or me for help. It will take some of your time and energy, but to save our children, future generations, and just maybe us from this curse, this is a petty price to pay.

To paraphrase Barry Goldwater's 1964 presidential nomination acceptance speech, "extremism in the defense of 'our children' is no vice. And let me also remind you that moderation in the pursuit of 'this goal' is no virtue."

—J. D. Griffith



## 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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## 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, 1D (EGR2), 1X, HNPP, 2E, 4E, and 4F can now be diagnosed by a blood test.
- ... is the focus of significant genetic research, bringing us closer to answering the CMT enigma.

## The CMTA Report

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



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