

SEPTEMBER 2003

The CMTA Report

Vol. 18, No. 4 ISBN #1067-0181

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.

Our Vision:

A world without CMT.

THE POWER OF ONE.. PLUS ONE...PLUS ONE

Individuals Working to Raise Funds for the CMTA

On Monday July 21, 2003, a golf tournament was held at The Creek, Locust Valley, Long Island, NY. Due primarily to the work of Board members Robert Kleinman, Phyllis Sanders, and Richard Sharpe, their fourth annual golf tournament netted \$38,000 for the research fund of the CMTA.

Approximately 60 golfers enjoyed the challenging golf course. Overcast and sometimes rainy skies gave way to a beautiful sunny, steamy day which made the event even more enjoyable. AFA Protective Systems, the company of which Robert Kleinman is Chairman, underwrites the event in order to allow more of the profits to benefit the organization.



Dwight White, former All-Pro Steeler, poses with tournament organizer Patrick Torchia (right) and Greg McNamara (left), a Florida participant at the Johnstown tournament.



Board member Phyllis Sanders, her husband, and daughter join Board Chairman Dick Sharpe in a break during the golf tournament at the Creek.

CMTA BOARD

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On Friday, July 25, 2003, the Johnstown area golf tournament took place at the Sunnehanna Country Club, home of the Sunnehanna Amateur Invitational, an event which began the careers of golfing greats Ben Crenshaw, Fred Couples, and Tiger Woods. Patrick Torchia, CMTA Board member, organized and ran this tournament entirely on his own and has done so for three years. Forty golfers, including Pittsburgh Steeler greats, Dwight White, L.C. Greenwood, and Robin Cole were in attendance. So far, his tournament has raised over \$13,000 for the CMT with some proceeds still outstanding.

(continued on page 2)

EMAIL CMTA AT:
CMTAssoc@aol.com



Betty Chow with her son Brandon, who has CMT and works at Trader Joe's business office. Trader Joe's donated a \$50 gift bag to the "Bowl Over CMT."

INDIVIDUALS RAISING FUNDS

(Continued from page 1)

A member of the CMTA, Betty Chow, with the help of her husband and son, staged a "Bowl Over CMT" on Saturday June 21, 2003 in Pasadena, CA. In addition to the bowling, Betty networked with other California CMT members and support groups and collected a total of \$3,145.65. Combined into that total was a \$1,000 matching gift from Wal-mart, which Betty's husband Henry was able to solicit. Serena Shaffer, former support group leader from Los Angeles, helped by gathering contributions from her co-workers and friends. Betty's efforts demonstrate what a grassroots fundraiser can do to help the CMTA. The total raised goes to the research fund of the organization.

The final individual raising money for CMT research is Lynn Upton of El Segundo, CA. Lynn will participate in a Walk 'n' Roll event

Frequently Asked Questions About Planned Giving

What is Planned Giving?

It is charitable giving that is planned, often deferred, and usually spread out over an extended period of time.

What is a Planned Gift?

It is a gift given to a charity through a will, contract, or trust.

What are some types of Planned Gifts?

Popular examples include life insurance policies and bequests of money, stocks, or tangible possessions. Gift annuities and life estates are more complex. Additionally, there are the very complex trust entities—charitable remainder trusts and lead trusts.

- **Will:** Legal document stating the disposition of a person's property upon his/her death.
- **Bequest:** Specific gift of property or assets.
- **Death Benefit:** proceeds from a life insurance policy's face value payable to the beneficiary.
- **Gift Annuity:** Contract by which a donor transfers cash or property in exchange for income payments for life.
- **Life Estate:** Donor gives home or land and retains rights to occupy or benefit from the property for life.

- **Charitable Remainder Trust:** A trust which returns income, fixed or variable, to the donor or others for life, or a period not to exceed 20 years.
- **Charitable Lead Trust:** A trust whereby the donor transfers assets/funds to a trust which, in turn, pays the charity for a certain period of time.

How can the CMTA support my planned giving efforts?

1. Our Executive Director can advise and assist prospective donors interested in making planned gifts for the benefit of the CMTA.
2. Our staff can assist the staff, volunteers, and estate planners who deal directly and confidentially with a prospective donor.
3. We can prepare customized gift illustrations that summarize the income and tax benefits of various forms of planned giving.
4. We can make presentations to staff and leadership volunteers interested in promoting planned giving for the benefit of the CMTA. ★

Interested in more information about planned giving? Contact Charles Hagins at 1-800-606-2682, option 6.



Wal-Mart's manager Sharon Rico with Henry Chow who asked for a matching gift from the retailer.

and is a team leader for CMT. She will have walked on Sunday, August 17th at the Mile Square Park in Fountain Valley. She solicited friends to walk with her or to support her as sponsors. Perhaps even more significantly, Lynn had her story publicized in the local paper, *The El Segundo News*. They wrote about her symptoms and the fact that the disease has not stopped her from having a successful career in the aerospace industry, a marriage, and children. But she told them that future medical advances could provide hope for her and others like her, so she wanted a way to help raise money for CMT research.

The Walk 'n' Roll event, organized by the Muscular Dystrophy Association, provided her with the perfect opportunity. To date, she has raised \$1,195 but hopes to increase that amount significantly. The one-mile walk may be more than Lynn can accomplish on her own and she may need to be pushed part of the way, but she'll make it to the finish line and in the process will demonstrate the "power of one." ★



Lynn Upton has solicited friends and family to "Walk 'n' Roll" for CMT.

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		\$40	
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	
CMT Facts V		active members \$12 inactive members \$15	
A Guide About Genetics for the CMT Patient <i>No shipping and handling on this item only.</i>		active members \$4 inactive members \$5	
Golf Shirt 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	
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 \$4.50</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 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.

Excerpt from Charcot-Marie-Tooth

By THOMAS D. BIRD, MD

EVALUATION STRATEGY

Establishing the specific cause of CMT hereditary neuropathy for a given patient involves a medical history, physical examination, neurologic examination, and nerve conduction velocity (NCV) and EMG testing, as well as a detailed family history and the use of DNA-based testing, when available.

CLINICAL EVALUATION

In individuals who have no family history of neuropathy, the first step is to exclude potential acquired causes of neuropathy by standard neurologic evaluation. In CMT1, the most common variety, NCVs are very slow and peripheral nerves may be palpably enlarged. This is not true of CMT2 or CMTX.

FAMILY HISTORY

A three-generation family history with attention to other relatives with neurologic signs and symptoms should be obtained. Documentation of relevant findings in relatives can be accomplished either through direct examination of those individuals or review of their medical records, including the results of molecular genetic testing and EMG and NCV studies. Patients with CMT may have a negative family history for many genetic reasons, including mild subclinical expression in other family members, autosomal recessive inheritance, and a new mutation for a dominant gene. About one third of patients with identifiable mutations causing the CMT1 hereditary neuropathy phenotype have new (*de novo*) mutations, and thus present as “sporadic” cases.

MOLECULAR GENETIC TESTING

Molecular genetic testing is clinically available for CMT1A, CMT1B, CMT1D, CMT2E, CMT4E and CMT4F, and CMTX. Because molecular genetic testing is available for mutations in several different genes associated with remarkable phenotypic overlap, the following strategy may provide the most efficient and cost-effective approach to testing. However, it should be noted that in many clinical laboratories, the testing for mutations involving hereditary neuropathy genes is done as a grouped panel, which may be less expensive than sequential testing of each individual gene (if more than two or three genes are analyzed).

TESTING STRATEGIES FOR PATIENTS WITH CMT

Positive family history.

In families with at least two-generation involvement, known male-to-male transmission, and slow NCVs, the CMT1A (*PMP22* dup) test should be obtained first, and then, if normal, followed by the CMT1B (*MPZ*) test.

In families with at least two-generation involvement and slow NCVs, but without male-to-male transmission, CMT1A, CMT1B, and CMTX DNA tests should be done sequentially.

In families with probable X-linked inheritance of the CMT phenotype, molecular genetic testing of the *GJB1* gene (encoding the protein connexin 32) for CMTX is appropriate in order to confirm the diagnosis.

In patients with the CMT2 phenotype, molecular genetic testing of *MPZ* and *GJB1* is appropriate, given that the CMT2 phenotype can be seen in patients with these mutations.

Negative family history.

CMT1A, CMT1B, and CMTX DNA tests should all be performed on males and females who have no family history of neuropathy, because new duplications of the 17p11 region often occur, giving rise to CMT1A, and because female carriers of a *GJB1* mutation causing CMTX may be asymptomatic.

Testing for rare causes of CMT.

Mutations in *EGR2* (CMT1D, CMT4E), *NFL* (CMT2E), and *PDX* (CMT4F), and point mutations in *PMP-22* are rare causes of the CMT phenotype. DNA-based tests are available to identify mutations in these genes. When tests for the more common forms of CMT are negative, the physician must decide if searching for the other, much more rare types of CMT justifies the cost. Prognosis and genetic counseling are frequently mentioned reasons for such extensive testing.

NEGATIVE MOLECULAR GENETIC TESTING RESULTS

Negative DNA testing results do not rule out a diagnosis of CMT since those normal test results are compatible with undetected mutations in other genes causing hereditary neuropathy.

Hereditary Neuropathy Overview

GENETIC COUNSELING ISSUES

Considerations in families with an apparent *de novo* (new) mutation. When the parents of a child with an autosomal or X-linked dominant condition are unaffected, possible non-medical explanations include alternate paternity or undisclosed adoption.

Family planning. The optimal time for determination of genetic risk, clarification of carrier status, and discussion of the availability of prenatal testing is before pregnancy. Similarly, decisions about testing to determine the genetic status of at-risk asymptomatic family members are best made before pregnancy. One study found that many patients with CMT give themselves high disability ratings and 36% would choose not to have children.

Testing of asymptomatic adult relatives who are at risk of developing CMT is possible after direct DNA testing has identified the specific gene mutation in an affected relative. Such testing should be performed in the context of formal genetic counseling.

Testing of asymptomatic at-risk children is discouraged. (See also the National Society of Genetic Counselors resolution on genetic testing of children).

DNA banking. DNA banking is the storage of DNA that has been extracted from white blood cells for possible future use. Because it is likely that testing methodologies and our understanding of genes, mutations, and diseases will improve in the future, consideration should be given to banking DNA. DNA banking is particularly important in situations in which molecular genetic testing is available on a research basis only or the sensitivity of currently available testing is less than 100%.

PRENATAL TESTING

Prenatal diagnosis for pregnancies at increased risk for CMT1A, CMT1B, CMT2E, or CMTX is possible. DNA extracted from cells obtained by chorionic villus sampling (CVS) at about 10 to

12 weeks' gestation* or amniocentesis at 16 to 18 weeks' gestation is analyzed. The disease-causing allele of an affected family member must be identified before prenatal testing can be performed.

Requests for prenatal diagnosis of (typically) adult-onset diseases are uncommon. Differences in perspective may exist among medical professionals and within families regarding the use of prenatal testing, particularly if the testing is being considered for the purpose of pregnancy termination rather than early diagnosis. Although most centers would consider decisions about prenatal testing to be the choice of the parents, careful discussion of these issues is appropriate.

MANAGEMENT

No treatment for CMT that reverses or slows the natural disease process exists. Treatment is symptomatic and patients are often evaluated and managed by a multi-disciplinary team that includes neurologists, physiatrists, orthopedic surgeons, and physical and occupational therapists. Daily heel cord stretching exercises to prevent Achilles' tendon shortening are desirable. Special shoes, including those with good ankle support, may be needed. Patients often require ankle/foot orthoses (AFOs) to correct foot drop and aid walking. Orthopedic surgery may be required to correct severe pes cavus deformity. Some patients require forearm crutches or canes for gait stability, but fewer than 5% of patients need wheelchairs. Obesity is to be avoided because it makes walking more difficult. Exercise is encouraged within the patient's capability and many individuals remain physically active. Important career and employment implications exist because of the persistent weakness of hands and/or feet.

Drugs and medications such as vincristine, paclitaxel, cisplatin, isoniazid, and nitrofurantoin that are known to cause nerve damage should be avoided.

The cause of any pain should be identified as accurately as possible. Musculoskeletal pain may respond to acetaminophen or nonsteroidal anti-inflammatory agents. Neuropathic pain may respond to tricyclic antidepressants or drugs such as carbamazepine or gabapentin. Initial, but not long-term improvement has been shown in a few patients with CMT1 and sudden deterioration who were treated with steroids (prednisone). ★

* Gestational age is expressed as menstrual weeks calculated either from the first day of the last normal menstrual period or by ultrasound measurements.

**The Charcot-Marie-Tooth Association
and
The University of Pittsburgh Medical Center, Shadyside
present
A One-Day Conference on CMT**

A conference on Charcot-Marie-Tooth Disorders will be held on Saturday, October 4, 2003, at the Herberman Conference Center, 5230 Centre Avenue, Pittsburgh PA, at the University of Pittsburgh Medical Center Campus.

The conference will begin with registration and continental breakfast from 8:00 to 9:00 am. Following registration, a welcome by the CMTA will be followed by an overview by Dr. Neil Busis, neurologist, on CMT. Athena Diagnostics will present a discussion on genetics and genetic testing for CMT. The final morning presentation will be on pain management. At noon, the attendees will enjoy lunch and the opportunity to speak personally with the doctors and other patients in attendance.

Following lunch, there will be presentations on surgery options, exercises, and bracing ideas. Recent research breakthroughs may be presented as well.

There will be time allotted for questions following each presentation. Directions to the hospital will be sent with confirmation of your registration. The capacity of the facility is 200.

PITTSBURGH CONFERENCE REGISTRATION FORM

Name _____

Address _____

(City) _____ (State) _____ (Zip) _____

Names of other attendees _____

Daytime phone number _____

Cost: \$35 per registrant with active CMTA membership **\$45 for non-current members**

Number of attendees: _____ Total amount _____ Check ☐ Visa ☐ MC ☐ AE ☐

Credit Card Number _____

Expiration Date _____ / _____ Signature _____

Please mail registration form and payment to:

Pittsburgh Conference
CMTA
2700 Chestnut Street
Chester, PA 19013

Or, return credit card payments by fax: 610-499-9267

Questions? Call 1-800-606-2682

Who Am I?

By Marianne DeStefano-Hill

(Author's Note: This essay was an assignment for a class I took last fall. We were to look into our past experiences and tell who we are now. I realized that when I'm wearing shorts and everybody can see I'm wearing braces, that makes me handicapped. I realized that I was so much more than someone with CMT, and I didn't want to be overlooked for all the other things I have done and am still able to do).

A little girl so pale and frail that I was called "Little Mary"
A child that loved to sing all day long until Mom said,
"No singing at the dinner table."

I fall down.

The only child out of six who would eat tomatoes
But could not seem to swallow chili.

The one who got straight A's in elementary school.

The little girl who liked to cook and craft things by hand,
But, I fell down.

The one who in high school was so filled with angst
That I retreated to my bedroom with the Beatles
And wrote songs to a world that would never understand.

I fell down.

A young woman who managed a book store...Heaven, really.
Every volume like a Christmas gift begging to be opened.
The smell of the new paper and the print of the text was intoxication.

And the words! Surrounded by millions and millions of words...
Knowledge is power.

I still fall down; I ache.

The journey to discover who I am continues
With more than a few men, and more than a few chemicals.
Searching, searching, searching for someone to understand me,
Know me, love me, tell me who I am.

I don't want to fall down anymore.

"It is genetic...progressive...neuromuscular disease
Charcot-Marie-Tooth"WHAT?!

It is not the end of the world.

I have a home, family, children, God....

So many blessings to be grateful for.

Music so beautiful it makes me cry to hear it.

Friends, flowers, chocolate!

Life is pretty good...I am managing.

Parenting is hard; parenting alone even harder.

I don't fall down much anymore.

My son falls sometimes now.

We have a bond between us

We know how it feels to be the last one picked for softball.

I recognize that I sound just like my Dad when I say,

"I can't understand what they're saying,"

When I try to listen to my son's music.

I recognize that my hands look just like my Mother's hands.

I notice when my brothers and sisters and I laugh,

We all sound the same.

So, who am I?

I am proud to be the daughter of Joseph and Margaret

Happy to be the mom of Nick and Shawn

Delighted to be a former Store Manager of the Year

Thrilled to be a student who just happens to enjoy singing
karaoke

A woman who wants to make a difference in the world....

Who is so much more than just a person with a disease.

One who garners recognition for her garden, her desserts,

Her painted "artsy crafts" items.

A woman who in her forties is still called

"Little Mary"

RADIO PERSONALITIES PROMOTE INFO ON CMT



"The God Squad" radio personalities, Rabbi Gellman and Father Hartman, attended the AFA Golf tournament. They have agreed to tape a show about CMT and other neuropathies on September 18, 2003.

GIFTS WERE MADE TO THE CMTA:

IN HONOR OF:

Alan Arcier

Richard Sharpe

Yohan Bouchard

Pierre & Mireille Aulagnier

Rima Gannage

Raphael & Joelle Gernez

Robert Chetlin

Honey & Carl Adelsheim

Marilyn & Charles Freed

Estelle & Melvin Mann

Bob & Ellen Selker

Lisa Curtis

A Friend

Richard Davis

Pat & Skip Davis

Mr. & Mrs. Charles Ratcliffe, Jr.

Sandra Ettelson

Nancy & Bob Fraiman

Mike & Joan Wald

Julie Leonard

Shirley Weiner

Paul Mobley

Douglas Mobley & Carolyn Rowley

Steve O'Donnell

West Chester CMT Support Group

Missy Post

Pamela & Anthony Osegueda

Charles Ratcliffe, Jr.

Mr. & Mrs. Richard Davis

Harold Seybert

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Leah Vaknin

Leon Gelman

Bernice & Ed Zeller

Julie Leonard

Judy Rosen & Family

Barbara & Steve Rothstein

Susi & Michael Sentlowitz

IN MEMORY OF:

Edith Aschenbach

Kay Flynn

John Basner

Franco & Virginia Camporesi

Steven Freedman

Cheri & Kevin Happenman

Sally & Herbert Kepner

Dr. & Mrs. Walter Krefetz

G. Jeffrey Machado

Donald & Joan Monson

Linda Rowley

Iona Samson

Robert & Charlotte Samson

Carol Smith

Marian & Chester Tyminski

Frances Chase

Louise Potocny

William Harder

Morris & Betty DePouw

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: _____



Cora Heard

Joann Adams
Ben & Kathleen Deane
Ray & Daffo Dooley
Dorothy Jones
Robert & Darlene Lonsinger
Elaine & Bill McMillin
Clarissa & Hershel Moore
Ferdinand & Esther Pall
Mr. & Mrs. Livio Palla
Dr. G. Parrott
Richard Rutherford
Vince & Lynn Schmitz
Maureen & Dan Sheffield
Frederick & Lee Valentich
Walt & Joyce Williams

Ruth Hom

Charles Hom

Dr. Louis Koenig

Leavern Hanson

Bertha Osborn

John & Glenda Ballentine
Maurice & Helen Benham
Robert Dalchow Family

Bertha Osborn (continued)

Rachel Davis
Ruth DeMott
Janet L. Dominique
Jim & Ginny Hull
Kathleen Hull
Norma Libby Hull
Dave & Ione Kaiser
Ken & Ruth Kaiser
Pamela Klingaman
Tom & Margaret Leach
Les & Jane Mallery
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Winnie Nowlin
Charles & Jacque Osborn, George & Megan
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Rolin & Rose Richardson
The Shanks
Char Taylor
Marion Thurow
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Elden & Shirley Winchell
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Seymour Ravinsky

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Minerva Slutzky & Jim Ebert

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Edward & Rosalie Silbert
Marc & Barbara Silbert

Ken Takayama

Ken's Friends from Bechtel
Mr. & Mrs. Kearny Chun
Mr. & Mrs. John Danton
Mr. & Mrs. Al Gee
Roland Kutaka
Winnie Lo
Mr. & Mrs. Tim Lum
Whitney Marr
Parsons/HNTB Tacoma Narrows
Ken's Friends @ Parsons Corp.
Diane Savard
Tacoma Narrows Construction
Patricia & Michael Wong

Make Your Voice Heard

The Individuals with Disabilities Education Act (IDEA) must be reauthorized this year. Parents of children with disabilities who are upset with the proposed revisions of existing law are angry that Congress has not listened to their concerns. They have formed a coalition and invite concerned parents to work with them on revising the law called National Committee of Parents Organized to Protect IDEA. Contact: Larry Searcy at 202-986-3000 or 202-487-4186.

For questions about IDEA visit www.ideapractices.org

To understand the original amendments from 1997, visit www.ed.gov/offices/OSERS/policy/IDEA

To see the new reauthorization concepts from the National Council on Disabilities, visit www.ncd.gov/newsroom/publications/synthesis_07-05-02 ★



■ A CHANCE TO TRY EASY GAIT

Easy Gait will be attending the World Conference on Disabilities in Orlando on September 18, 2003, where 7,000 to 8,000 people are expected. They will measure attendees for braces on the spot and give them an opportunity to walk around the Convention in them to determine if they like the fit and feel of the brace before committing to purchase it. Keith Ahern, a ten-year old boy with CMT from Ft. Lauderdale will be their "poster boy" at the booth and show people how well he can walk in Easy Gait.



Suggestions for Managing Your Disease

(Ideas from the Mayo Clinic.)

Take care of your feet. Tight shoes and socks can worsen pain and tingling, and may lead to sores that won't heal. Wear soft, loose cotton socks and padded shoes. You can use a semicircular hoop, which is available in medical supply stores, to keep bedcovers off sensitive or hot feet.

Soak your feet or hands in cold water. If you have burning pain, cool off your feet or hands in cold, but not icy, water for 15 minutes twice a day. This is particularly useful at night. After soaking, rub on petroleum jelly to soften your skin.

Exercise. Ask your doctor about an exercise routine that's right for you. Regular exercise may reduce neuropathic pain and can help maintain unaffected muscles.

Reduce your consumption of alcohol. There is a danger in over-consumption of alcohol because of the risk of falls and the fact that alcohol can worsen any neuropathy.

Limit caffeine. Try to avoid foods or drinks high in caffeine, including coffee, chocolate, and sodas. Caffeine can worsen pain.

Eat healthy meals. Emphasize low-fat meats and dairy products and include lots of fruits, vegetables, and whole grains in your diet.

Quit smoking. Smoking can worsen symptoms. If you smoke, talk to your doctor about ways to quit.

Massage your hands and feet. Or, have someone massage them for you. Massage helps improve circulation, stimulates nerves, and may temporarily relieve pain.

Coping Skills

Living with chronic pain or disability presents daily challenges. Some of these suggestions may make it easier for you to cope:

Set priorities. Decide which tasks you need to do on a given day and which can wait for another time. Stay active, but don't overdo.

Get out of the house. When you have severe pain, it's natural to want to be alone. But this only makes it easier to focus on your pain. Instead, visit a friend, go to a movie, or take a short walk.



Regular exercise may reduce neuropathic pain and help maintain unaffected muscles.

Seek and accept support. It isn't a sign of weakness to ask for or accept help when you need it. In addition to support from family and friends, consider joining a support group for CMT patients or for neuropathy or chronic pain. Support groups aren't for everyone, but they are good places to hear about coping techniques or treatments that have worked for others. You'll also meet people who understand what you are going through.

Prepare for challenging situations. If something especially stressful is coming up in your life, such as a move or a new job, knowing what you have to do ahead of time can help you cope.

Talk to a counselor or therapist. Insomnia, depression, and impotence are all possible complications of peripheral neuropathy. If you experience any of these, you may find it helpful to talk to a counselor or therapist in addition to your primary care doctor. There are treatments for each of these conditions that can help. ★

The Invisible Disease: Depression

(from National Institute of Mental Health)

Depression is a serious medical condition. In contrast to the normal emotional experiences of sadness, loss, or passing mood states, clinical depression is persistent and can interfere significantly with an individual's ability to function. There are three main types of depressive disorders: major depressive disorder, dysthymic disorder, and bipolar disorder (manic-depressive illness).

SYMPTOMS AND TYPES OF DEPRESSION

Symptoms of depression include sad mood, loss of interest or pleasure in activities that were once enjoyed, change in appetite or weight, difficulty sleeping or oversleeping, physical slowing or agitation, energy loss, feelings of worthlessness or inappropriate guilt, difficulty thinking or concentrating, and recurrent thoughts of death or suicide. A diagnosis of major depressive disorder is made if a person has five or more of these symptoms and impairment in usual functioning nearly every day during the same two-week period. Major depression often begins between ages 15 to 30, but also can appear in children. Episodes typically recur.

Some people have a chronic but less severe form of depression, called dysthymic disorder, which is diagnosed when depressed mood persists for at least 2 years (1 year in children) and is accompanied by at least 2 other symptoms of depression. Many people with dysthymia develop major depressive episodes.

Episodes of depression also occur in people with bipolar disorder. In this disorder, depression alternates with mania, which is characterized by abnormally and persistently elevated mood or irritability and symptoms including overly-inflated self-esteem, decreased need for sleep, increased talkativeness, racing thoughts, distractibility, physical agitation, and excessive risk taking. Because bipolar disorder requires different treatment than major depressive disorder or dysthymia, obtaining an accurate diagnosis is extremely important.

FACTS ABOUT DEPRESSION

- Major depression is the leading cause of disability in the U.S. and worldwide.
- Depressive disorders affect an estimated 9.5 percent of adult Americans ages 18 and over

in a given year, or about 18.8 million people in 1998.

- Nearly twice as many women (12 percent) than men (7 percent) are affected by a depressive disorder each year.

Depression can be devastating to family relationships, friendships, and the ability to work or go to school. Many people still believe that the emotional symptoms caused by depression are “not real”, and that a person should be able to shake off the symptoms. Because of these inaccurate beliefs, people with depression either may not recognize that they have a treatable disorder or may be discouraged from seeking or staying on treatment due to feelings of shame and stigma. Too often, untreated or inadequately treated depression is associated with suicide.

RESEARCH FINDINGS

- Brain imaging research is revealing that in depression, neural circuits responsible for moods, thinking, sleep, appetite, and behavior fail to function properly, and that the regulation of critical neurotransmitters is impaired.
- Genetics research, including studies of twins, indicates that genes play a role in depression. Vulnerability to depression appears to result from the influence of multiple genes acting together with environmental factors.
- Other research has shown that stressful life events, particularly in the form of loss such as the death of a close family member, may trigger major depression in susceptible individuals.
- The hypothalamic-pituitary-adrenal (HPA) axis, the hormonal system that regulates the body's response to stress, is overactive in many people with depression. Research findings suggest that persistent over-activation of this system may lay the groundwork for depression.
- Studies of brain chemistry, the action of antidepressant medications, and the cognitive distortions and disturbed interpersonal relationships commonly associated with depression continue to lead to the development of new and better treatments. ★

■ REACHING "HIGHER GROUND"

The Reverend Jim Ries, diagnosed with CMT as a teenager, has struggled with it his whole life. He has been a Catholic priest for the last ten years and was recently named pastor of St. Mary's Catholic Church in Orange County, California. In a local paper's story about his appointment, the writer said, "Ries won't be hard to spot as he makes his way to the altar. Born with Charcot-Marie-Tooth disease, the most common inherited neuropathy, he walks with a crutch to stabilize an awkward gait. Yet, even more evident are his sky-blue eyes that appear to laugh above his contagious smile." Congratulations to Jim!

Support Group News

■ Minnesota—Twin Cities

The first meeting of the Minnesota—Twin Cities CMT/HNPP support group will be held on Saturday, September 27, 2003 from 10 AM until noon. Dr. Gareth Parry will be on hand to speak about CMT and HNPP and to answer questions. For more information, please contact Maureen Horton at 651-690-2709 or email at mphorton@qwest.net. Bill Miller will be the co-chair and can be reached at 763-560-6654 or wmiller7@msn.com. If you can attend, please contact one of the leaders for location information.

■ Missouri—Eastern Kansas

The Kansas City CMT Support group, with co-hosts MDA and Kansas University Medical Center, Neurology Department, has scheduled an all-day workshop for Saturday, October 11, 2003, at the Landon Center, KU Medical Center, Kansas City, KS. In addition to discussion of the many points of interest for those with CMT, there will also be a special two-hour closed session for children, giving them time to talk to each other about problems unique to kids with CMT. There will be a pediatric neurologist and a pedorthist there and a resource person to address ADA (Americans with Disabilities Act), as this law affects school children.

As there is limited seating available, local members and guests will be given first preference. There are a number of people living in Eastern Kansas, Western Missouri and the borders of Nebraska and Iowa in touch with the local group. For information, email ardi5@aol.com or rushfeltj@aol.com or call 816-229-2614.

■ Missouri—St. Louis

The group met on Saturday, August 16, 2003, at the St. Louis Medical Center Cafeteria from 10AM to noon. The speaker was David Osterman, board-certified pedorthist and orthotist. For more information, contact Carole Haislip at 314-644-1664.

■ New York (Westchester County)/ Connecticut (Fairfield)

On June 21, 2003, the support group had their annual pot-luck brunch. Their leader, Beverly Wurzel says, "A good time was had by all."

The group sends their best wishes to Diane and Stan Kosik who are moving to Arizona.

Diane co-chaired the group and she and Stan helped out at many MDA events. The group will miss their company and their contributions.

The next meeting will be September 20, 2003. Dr. Anne Hanley, a neurologist from Albert Einstein, will be the guest speaker. CMT patients, family, and friends are always welcome.

■ Ohio—Northwestern Region

The group met for the first time on Saturday, July 26, 2003. The first meeting was a get-acquainted meeting to share information and to plan a fall meeting. Six participants attended. There was a lot of sharing and discussion about ankle-foot orthotics and management tricks and techniques that people have tried or discovered. William Lloyd displayed his toe-padding technique using cotton-ball "donuts" slipped over the toes to help with some of the symptoms he experiences. A mother and daughter discussed both the advantages and disadvantages of wearing AFOs. Our next meeting will be in October.

■ Pennsylvania—Northwestern Area

The group has 15 regulars with CMT and a few who have come to one or two meetings. The next meeting will be September 27th and representatives of Walk-Right, Inc. will be discussing their walking aids.

Typically, a packet of information is provided for members to take home, including articles like research developments, stories about others with CMT, medical issues concerning CMT, and sometimes a scientific article which has been translated into layman's terms so it's easier to understand. A page of internet links with websites that have helpful information for people with CMT—things like adaptive gadgets, extra-width and depth shoes, exercise equipment for home use, and social services is also included. At every meeting, the leader provides membership forms for the CMTA.

At the last meeting, leader Joyce Steinkamp, spoke at some length about her recent visit to the Wayne State CMT clinic in Detroit and answered questions about the testing. She encouraged members to visit the clinic themselves and if they could not, to at least participate in the North American Database by mail. ★

CMT Support Groups

Bob Budde, Support Group Liaison, 859-255-7471

Arkansas—Northwest Area

Place: Varies, Call for locations
Meeting: Quarterly. Meetings are not regularly scheduled so call ahead.
Contact: Libby Bond, 479-787-6115
E-mail: charnicoma57@yahoo.com

California—Berkeley Area

Place: Albany Library, Albany, CA
Meeting: Quarterly
Contact: Gail Whitehouse
E-mail: gwhite@earthlink.net

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: pcmoble@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, 502-695-3338
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

Minnesota—Benson

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

Mississippi/Louisiana

Place: Baptist Healthplex, 102 Clinton Parkway, 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: Horseheads Free Library on Main Street, Horseheads, NY
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: DianeK319@optonline.net 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—Centerville/Cincinnati

Place: Church of the Brethren
Meeting: Fourth Thursday, April–October
Contact: Shirley Planet, 937-433-1296

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—Johnstown Area

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

Pennsylvania—Northwestern Area

Place: Blasco Memorial Library
Meeting: Call for information
Contact: Joyce Steinkamp, 814-833-8495
E-mail: joyceanns@adelphia.net

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—State College

Place: Centre County Senior Center
Meeting: Call for information
Contact: Katia Skovrinskie, 717-608-1081
E-mail: skov@psu.edu

Dietary Supplements under Scrutiny Again

(From NORD On-Line Bulletin, July 2003)

All the bad news about dietary supplements is affecting sales. Americans are learning that the products are not tested for safety or effectiveness before they reach the market, and many people have been harmed.

Lawsuits against the supplement manufacturers are often settled out of court and therefore, the public is not ordinarily privy to the evidence against manufacturers. However, some recent class action suits have been filed against manufacturers on the grounds of false advertising, and some of the evidence is now a matter of public record, particularly in cases of ephedra (which was often used in diet remedies until the recent death of a Baltimore Orioles pitcher).

Supplement manufacturers like to advertise that their products are "proven effective" in "clinical trials," but in at least three recent cases against manufacturers of weight loss remedies judges found that study results have been fudged, and the science is often questionable.

Since there is no regulatory agency requiring decent scientific studies of supplements, a manufacturer can do a clinical trial of only 10 or 12 people, claiming that the results show the product is "clinically proven." However, in class action cases, correspondence, experimental records and emails were subpoenaed to show how companies suppressed negative data, removed product

names from negative abstracts, changed statistical methods to show improved results, and omitted patients who had side effects and dropped out of the experiments. They also showed how companies tried to intimidate investigators to change study results, and purposely misinterpreted data to use for their advertising campaigns.

Negative studies are often "buried" in supplement trials, according to one consultant. In one study, when some research subjects experienced an irregular heartbeat and elevated blood pressure from a nutritional supplement, the drug was ultimately advertised as "safe" because data from these drop-outs was omitted. In another study, several of the more seriously overweight subjects dropped out of the study but the statistician left their data in to calculate the average weight of people in the study, even though they were omitted from calculations at the end of the study. Thus, the company could exaggerate the appearance of weight loss.

The judge in a California case decided that the advertising claims were false and misleading because you cannot "pick and chose the data which would give the most favorable results" for a product. As more of this chicanery is exposed in class action lawsuits, the more public trust will be lost for the supplement industry. ★

CMTA Members Earn Research Match

The research appeal for 2003 has raised over \$45,000 to earn the \$30,000 match which was established by Gary and Vicky Gallagher. With their encouragement, by way of the challenge, the Gallaghers convinced 535 of you to give to the research appeal. We appreciate each and every gift.

In addition to the appeal, Steve O'Donnell's Swim for the Cure raised an additional \$51,000 and the AFA Golf Tournament raised \$38,000.

Even these excellent fundraisers were not able to get us to our goal of \$250,000. If you haven't yet contributed to the research fund, consider making a gift before the end of this year to make our goal a reality.



Steve O'Donnell's two swims have raised more than \$121,000 for CMT research.

Ask the Doctor

Dear Doctor,

I am 47 years old and was diagnosed with CMT 24 years ago. Both of my children have obvious CMT, while in my case, it is not apparent. I had an adrenal tumor removed three years ago and since then, subtle changes are evident in my hand function. Could removing the tumor have caused the progression? I also have problems with my kidneys and am removing too much protein through my urine. This makes me fatigued all the time. Is the kidney disease associated with CMT?

A Doctor from the Medical Advisory Board replies:

There have been occasional cases of patients with CMT having kidney disease, but there is no clear correlation between the two. Similarly, I am unaware of why removing an adrenal tumor would affect your CMT. However, significant renal abnormalities with high BUN and creatinine levels can cause neuropathy independent of CMT, and this could lead to an exacerbation of CMT. If the problem is mostly in your hands, you should be evaluated for carpal tunnel syndrome, which can occur in CMT and kidney disease.

■

Dear Doctor:

My son has CMT and is working as an emergency medical technician. He might be required to have a smallpox vaccination in order to keep his job. Do people with CMT have an increased risk of problems from such a vaccination?

A doctor replies:

CMT is not an autoimmune disease, so there is no theoretical reason why a patient with CMT could not have the immunization/vaccination. However, it should be confirmed that your son actually has CMT and not an autoimmune disease before proceeding with the vaccination.

■

Dear Doctor,

Hip replacement surgery is often a boon to those who suffer from arthritic joints and there is no other way to get relief. The operation, in itself, in the hands of a skilled surgeon, usually goes smoothly. The complication arises when the surgery is performed on people with CMT who have a tendency to fall when walking unaided or when using a cane.

Orthotics help, but are no protection against falling. I had my second hip replacement and am experiencing many more falls than I did with my first one. The obvious answer is to use a walker, which is much safer and more secure, but my objective is to get back to normal with no cane or occasional use of the cane. If I take the suggestion of my surgeon, it is to get back on the cane as quickly as possible to preserve my prior-to-surgery level of mobility. My therapist, concerned about my falling, wants me to spend more time on the walker. How would you solve this problem?

An Orthopaedic Surgeon replies:

It would be my understanding that the surgeon has probably done his/her job and the joint replacement is stable and pain-free. It is now up to the individual to improve his function. Falling is not good and, when uncontrolled, could lead to the dislocation of the hip and a major complication.

Age, balance, shakiness, memory, patience, shortness of breath, shoes, terrain, and obstructions are all important factors. The therapist should do a manual muscle test and determine just where the weakness is. Also, a functional evaluation of the person's surroundings, his/her home, yard, furniture placements, exercise, etc. is important. A therapist who has an interest in rehabilitation would be able to use this information and design a program to accomplish a reasonable, accomplishable (realistic), and mutually agreed upon goal. In this case, I believe it is to prevent falling and to go back to walking some distance without aids.

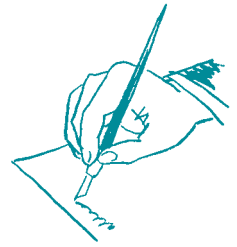
Knowledge of where the weakness is in an elderly, active, ambulatory person with long-standing manifestations of CMT could allow the therapist to develop a strengthening program for these muscles—for instance, the abdominals, the hip extensors, hip abductors, and quads and, together with lightweight AFOs, allow a person to balance better and walk with minimal aids.

Dear Doctor,

Could edema be a symptom of CMT?

A doctor replies:

Many people with CMT develop length-dependent edema (in feet) as they get older. It is usually treated with diuretics, foot elevation, and compression stockings.



WRITE TO US!

Pat Dreibelbis, Editor

The CMTA Report

CMTA

2700 Chestnut Pkwy.

Chester, PA 19013

The CMTA reserves the right to edit letters for space.

The CMTA Report

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

Photos on pages 9 and 10 by Ian Britton, courtesy of FreeFoto.com

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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