1983-2003
CELEBRATING 20 YEARS OF LIVING OUR MISSION

Finding a Cure | Creating Awareness | Improving Quality of Life

The CMTA Report

NOVEMBER 2003

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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.

CMTA BOARD

EMAIL CMTA AT:
CMTAssoc@aol.com

CMT Recognized in Two Media Events in Johnstown, PA

By J.D. GRIFFITH

Academy Award-winning actress Olympia Dukakis joined officials from the Johnstown-based Conemaugh Health System (CHS) to launch the John P. Murtha Neuroscience Institute, one of the few programs in the country that will blend advanced medical practices and research with time-honored healing methods to treat conditions related to the brain and nervous system, including CMT. Construction on the project begins soon and will be completed by late spring/early summer 2004. The permanent institute will be built in CHS's planned technology park.

The gala, on August 28, 2003, included a keynote address by Olympia Dukakis and talks by US Congressman John Murtha, Pennsylvania Lieutenant Governor Catherine Baker Knoll, and others. Ms. Dukakis applauded CHS, its national partners, and Congressman Murtha for taking the initiative to expand neuroscience care and bring the latest treatments and methods to west central Pennyslvania. "Neurological disease knows no boundaries," she said. "It can affect anyone, anytime."

Charles F. Hagins, CMTA Executive Director, and I were among the invited guests. I have never heard Charcot-Marie-Tooth mentioned, albeit occasionally mispronounced, so often at a public forum. It send chills up my spine.

In addition to advanced medical technologies located at the Institute, the programs will use nondrug treatment methods to help patients manage and control their symptoms as well as any pain related to their medical condition. National clinical studies, developed in partnership with Walter Reed Army Medical Center, the National Naval Medical Center, and the Uniformed Services University of the Health Sciences, will also be conducted at the site. Through these and other partnerships, patients will have direct access to the latest findings in neuroscience care.

Olympia Dukakis and Senator John Murtha discuss the potential for the new Neuroscience Institute in Johnstown, PA.

A second CMT media event, proclaiming September as CMT Awareness Month, was held in the Atrium of the Conemaugh Hospital on Franklin Street in Johnstown, PA. The local television channels covered the event, with spots on the 6:00 and 11:00 PM news. CMT Awareness

(continued on page 3)

FISCAL YEAR JULY 1, 2002 TO JUNE 30, 2003

Annual Report

By CHARLES F. HAGINS, Executive Director

This has been another successful year of growth and development for the CMTA. The post-9/11 economy certainly created a challenging environment in which non-profits had to try to raise operating and research funds. However, with the efforts of our dedicated staff we were able to keep operational expenses under budget and maintain giving levels for both the Operations' Appeal and Research Appeal.

Fundraising events, sponsored by members of the Board of Directors and others, continue to be important sources of revenue for the CMTA. Three golf tournaments and a swim produced approximately one-third of our fundraising income. The development efforts of the Executive Director and staff created another one-third of the revenue allocated toward operations and research.

The Board of Directors added a new nonvoting member, Amar Kamath. Mr. Kamath is employed by Athena Diagnostics. He holds an MBA degree and has vast experience in marketing and sales. His knowledge of CMT and other neuropathies will enable him to guide the strategy of the CMTA as it pertains to the research community and the members we serve.

The CMTA joined forces through a defined collaboration with Conemaugh Health System of Johnstown, PA to help outline the Best Practice Guidelines for the newest CMT Clinic in the country at the new Regional Neuroscience Center, which is located in Johnstown, PA. The efforts of Congressman John P. Murtha (D) helped make this collaboration possible.

In addition, the CMTA attended the conventions of both the American Neurological Association in New York City and the American Society of Human Genetics in Baltimore, Maryland.

The CMT Support Group network continues to grow throughout the country and creates a network for volunteers to share information and advances in research at a grassroots level. The CMTA has no financial obligation to the Support Group network; however, we do support them with timely information about the disease and we continue to urge participants to join the CMTA and receive our direct benefits of membership.

The CMTA was extremely pleased with the success of the First Annual Symposium of the North American CMT Consortium held in March 2003 in London Ontario. Seventy scientists presented results from their research and participated in the three-day networking event. It was agreed at this meeting that the event would rotate every other year between North America and Europe. Next year's event is scheduled for July 2004 in Antwerp, Belgium.

The CMTA plans to open dialogue with the National Institutes of Health to secure increased funding for CMT research.

Strategically the Board of Directors has decided to make a focused effort to open dialogue with the National Institutes of Health (NIH) to share ongoing communication regarding the issues and efforts of the CMTA, to understand how to utilize the NIH, and ultimately to secure increased funding for research for CMT. The CMTA believes that NIH is a significant source of research funding that would be difficult to match in the private sector through our own efforts and that this disease deserves its rightful share of these tax dollars.

As we celebrate the CMTA's 20th anniversary and reflect on our beginning, I believe we have much to celebrate:

- 3,000 current members
- CMTA North American Database
- First Annual Symposium of North American CMT Consortium
- 25 support groups
- Collaboration with Conemaugh Health System's new Regional Neuroscience Center, CMT Clinic
- Collaboration with National Institutes of Health

We remain dedicated to 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 disorders." ★

TWO EVENTS

(Continued from page 1)

Month was proclaimed in Johnstown, PA, by Mayor Don Zucco and in the State of Pennsylvania by State Senator John Wozniak. A plaque was presented to me, in recognition of my work as the Johnstown CMT support group leader. The Cambria County Commissioners were also in attendance. A statement from US Congressman, John P. Murtha, was also read by a representative of his office. The following is an excerpt from that statement:

"CMT has received too little attention for far too long. Today's event, to highlight CMT Awareness Month, is one step in the right direction…CMT is the most common inherited neurological disorder, affecting 150,000 Americans, and it can be severely debilitating or, in some cases, even prove to be fatal. With such an impact, I hope we can make more people aware of this devastating disease, improve the potential for prompt diagnosis, and improve the treatment protocol to reduce or slow down the impact on those who suffer from the disease."

Congressman Murtha, a 29-year member of Congress, has been very generous in securing funding for health care. In a conversation with him, he said, "Anything I can do for CMT, please ask." We will!

Please help with CMT Awareness Month 2004. Contact your elected officials: mayors, state and national legislators, governors, the President. They all have a public e-mail, an address, or a phone number. Plead your case. They are generally happy to help; they get recognition, publicity, votes, and a warm fuzzy feeling. Plan an event in your area for next September. ★

J.D. Griffith was presented with a plaque recognizing his contribution to CMT Awareness Day.

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 ☐ English ☐ Spanish		*active members* $3 ***inactive members* $5**	
CMT Facts II ☐ English ☐ 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 *No shipping and handling on this item only.*		*active members* $4 ***inactive members* $5**	
Golf Shirt *Size:* ☐ M ☐ L ☐ XL ☐ XXL		$15	
CMT Informational Brochure ☐ English ☐ Spanish		FREE	
Physician Referral List: States: _____ _____ _____		FREE	
Letter to Medical Professional with Drug List		FREE	
Contribution to CMT Research *10% will be applied to administrative expenses.*			
Shipping & Handling *Orders under $10 add $1.50, orders $10 and over add $4.50*			
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.

2003–2004 Grants that Focus on CMT Research

(Editor's note: In addition to the five grants that the CMTA is funding this year, there are numerous grants funded by the Muscular Dystrophy Association that focus on CMT-related issues. We present this list for you to demonstrate that research into the causes and possible cures for CMT is an ongoing process.)

(RG)= Research grants
(DG)= Development grants

CALIFORNIA

Stanford University

Eric Shooter, Ph.D.
(RG) Neurotrophin therapy for Charcot-Marie-Tooth disease (CMT)
(RG) Peripheral myelin protein 22 (PMP22) in Charcot-Marie-Tooth disease type 1 (CMT1)

COLORADO (BOULDER)

University of Colorado

Kevin R. Jones, Ph.D.
(RG) Neurotrophic factor requirements in peripheral nerve

FLORIDA (GAINESVILLE)

University of Florida

Lucia Notterpek, Ph.D.
(RG) Targets for therapy in the cellular pathogenesis of Charcot-Marie-Tooth (CMT) neuropathies

MASSACHUSETTS

Massachusetts General Hospital

Khemissa Bejaoui, Ph.D.
(DG) Cloning and characterization of the HSN-1 gene

MICHIGAN

Wayne State University

Michael Shy, M.D.
(RG) Adeno associate virus gene therapy for Charcot-Marie-Tooth (CMT)

PENNSYLVANIA

University of Pennsylvania

Christiane Massicotte, D.V.M., M.S., Ph.D.
(RG) Trafficking of Cx32 mutant protein that cause inherited neuropathy

TEXAS

Baylor College of Medicine

Ken Inoue, M.D., Ph.D.
(DG) SOX10 and EGR2 transcription pathways may regulate genes involved in Charcot-Marie-Tooth (CMT)

James Lupski, M.D., Ph.D.
(RG) Molecular genetics of Charcot-Marie-Tooth (CMT)

Pragna Patel, Ph.D.
(RG) Regulation of PMP22, the gene underlying CMT1A and HNPP

George Snipes, M.D., Ph.D.
(RG) Cellular biology of Charcot-Marie-Tooth (CMT) related disorders

WASHINGTON

University of Washington

Phillip Chance, M.D.
(RG) Gene isolation in families with Charcot-Marie-Tooth type 1C (CMT1C)

Valerie Street, Ph.D.
(DG) Gene isolation in families with Charcot-Marie-Tooth neuropathy type 1C (CMT1C)

WISCONSON

University of Wisconsin

John Svaren, Ph.D.
(RG) The role of EGR2 dysfunction in peripheral neuropathies

AUSTRALIA

University of Sydney

Garth Nicholson, Ph.D.
(RG) Construction and characterization of hereditary sensory neuropathy type 1 transgenic mouse

University of Western Australia

Luba Kalaydjieva, M.D., Ph.D.
(RG) Cloning the gene for a severe autosomal recessive form of Charcot-Marie-Tooth (CMT) disease

CYPRUS

Cyprus Institute of Neurology and Genetics

Kyproula Christodoulou, Ph.D.
(RG) Identification of a novel Charcot-Marie-Tooth Type 2 (CMT2) gene
(RG) Neuromuscular diseases in Eastern Mediterranean countries

Scarduzio Family Enjoys Golf Fundraising Success

On September 8, 2003, the John J. Scarduzio Memorial Golf Tournament was played at Rolling Green Golf Club, Springfield, PA. Ninety-eight golfers enjoyed a beautiful course and a gorgeous late summer day.

The entire day was a testament to the commitment and organization of the extended Scarduzio family. Lead by Christopher Scarduzio, the family solicited attendees, got hole-in-one prizes, prepared trays of homemade Italian cookies and candies for the silent auction, and showed up in force to play golf and do other volunteer activities. One family member donated the breakfast of eggs Benedict, fresh fruit, and pastries. Another donated the barbecue, which featured hamburgers, hot dogs, steaks, boneless chicken, and side dishes of endless variety. Because of the entire family's commitment to this event, the gross for the day was over $60,000.00. That income was significantly higher than in preceding years.

Michael Kirby contributed $15,000 in support of the research appeal. Pictured with him is John DeStefano

Among the attendees were Philadelphia 76er Todd MacCulloch and noted chef and owner of Le Bec Fin, Georges Perrier. Executive Director Charles F. Hagins and Board Chairman Richard L. Sharpe also attended and golfed to show their support for the fundraising effort. Each foursome received a picture of their group as a memento of the day.

While many of the people attending joked about the number of "Scarduzios" there were and how saying the name John, Bobby, Bernadette, or Mary Ann was sure to get numerous responses, the reality of the day was that the family was totally immersed in the effort and that every single family member was there in some capacity helping to make this the best event yet. This golf tournament showed the power of one family. . . but the support of many individuals. ★

The foursome of Philadelphia 76er Todd MacCulloch, Georges Perrier, Chris Scarduzio, and Chris Wallace (rear) posed at the end of the sixth hole at the Rolling Green Country Club.

Registering golfers, selling 50/50 tickets, and generally making sure things ran smoothly were Scarduzio family members: Darlene Scarduzio, Mary Ann Lucidi, and Roma Scarduzio.

TAX BREAKS FOR 2003

Year-End Gift Ideas

When you consider your year-end tax planning, we hope you will consider making use of the income tax charitable deduction. Your 2003 year-end gift can significantly reduce your income taxes, while providing meaningful support for the CMTA.

If you itemize on your tax return, you can almost always lower your income taxes through charitable giving. The amount of your savings will depend, of course, on your income tax bracket. For example, if you are in the 33% tax bracket for 2003, and you itemize, a $1,000 gift to the CMTA by December 31st will save you $330 in taxes.

Giving is, of course, much more than tax brackets and charitable deductions. Philanthropy provides the meaningful difference in what activities the CMTA can fund.

There is no easier way to garner a charitable deduction for 2003 and support us at the same time…than by simply writing a check. Make sure your envelope is postmarked by December 31st and your gift will qualify as a 2003 gift, even if we do not receive it until the first week in January. If you itemize, your outright gifts of cash are fully deductible for federal income tax purposes up to 50% of your adjusted gross income.

Another equally good way to give at the end of year is by making a gift of stock. First, in giving long-term appreciated stock, you avoid paying any capital gains tax on the increased value of your stock. Additionally, you receive a tax deduction for the full fair-market value of the stock on the date of the gift. For income tax purposes, the value of such gifts may be deducted up to 30% of your adjusted gross income.

If you are interested in making a different gift of property, please contact Executive Director Charles Hagins for advice on how to proceed with that gift. Always check with your accountant, tax attorney, or other tax advisor for additional information on how these general rules apply to your personal situation. ★

The God Squad Focuses on CMT

On Thursday, September 18, 2003, Dr. Fran Dyro, New York Medical College, Valhalla, NY, and a member of the CMTA's Medical Advisory Board and actress Nicolle Cyrille (previously profiled in *The CMTA Report*) were featured on a taping of *The God Squad*. The show is a production of Telecare and airs on local cable stations in the New York tri-state area with a concentration on Long Island. Telecare estimates that it reaches an audience of over 2 million viewers on Long Island alone.

Dr. Dyro and Nicolle discussed Charcot-Marie-Tooth disorders from the view of a professional who treats patients with CMT and the vantage point of a patient living with the problem day to day. The hosts, Rabbi Marc Gellman and Monsignor Thomas Hartman have frequently appeared on the radio (The Don Imus Show) and on national television (Good Morning America) and write a nationally syndicated weekly newspaper column.

The program on CMT was suggested by Board member Robert Kleinman and the arrangements for the taping were made by Mr. Kleinman and fellow Board member Dick Sharpe. The program is expected to air sometime in mid to late October. ★

Nicolle Cyrille, Rabbi Marc Gellman, Monsignor Thomas Hartman, and Dr. Fran Dyro appeared together on The God Squad, *airing on the Telecare Network.*

Clinical Features of Charcot-Marie-Tooth Disease

By MICHAEL SHY, M.D.
Center for Molecular Medicine and Genetics
Wayne State University School of Medicine
Detroit, MI

(This is an excerpt from a presentation by Dr. Shy, which was given at a professional gathering, entitled "Inherited Peripheral Neuropathies".)

DEJERINE-SOTTAS DISEASE (DSD)

Dejerine Sottas disease was classified by Dyck and Lambert to identify patients with severe disability beginning in infancy who had an autosomal recessive inheritance pattern. Subsequently, it has been shown that many presumed DSD patients have autosomal dominant mutations caused by mutations in PMP22, MPZ, and EGR2. We use DSD to define all patients with severe onset in infancy. Specifically, 1) onset by 2 years of age with delayed motor milestones, or 2) severe motor, sensory, and skeletal deficits with frequent extension to proximal muscles, sensory ataxia, and scoliosis.

CMT1A

Most CMT1 patients (85%) become symptomatic clinically in their first two decades of life. The largest group, CMT1A, usually develop a "typical" CMT phenotype. They are slow runners in childhood, develop foot problems in their teenage years, and often require orthotics for ankle support as adults. Variable degrees of hand weakness occur, typically lagging about ten years behind the development of foot weakness. Sensory loss, also variable, occurs in both large (vibration and proprioception*) and small (pain and temperature) modalities. While the combination of weak ankles and decreased proprioception often leads to problems with balance, the vast majority of patients remain ambulatory throughout their life, which is not shortened by their disease. Almost all patients with CMT1A have absent deep tendon reflexes. Most have foot deformities with high arches and hammer toes. One may be able to palpate the enlarged nerve trunks in subcutaneous tissue. Additional features, including postural tremor (referred to as Roussy-Levy syndrome) and muscle cramps, may also occur. While this phenotype is typical for CMT1A patients, it is not invariable. Occasional patients develop a severe phenotype in infancy, while others develop minimal disability throughout life. Since phenotypic variability occurs within the same generation within the same family, it is not yet possible to predict who will have more disabling forms of the disease.

HNPP

HNPP is caused by a deletion of the same region of chromosome 17 that is duplicated in CMT1A. HNPP patients present with a variety of patterns, which typically present as focal symptoms of weakness or sensory loss following compression of individual nerves. These symptoms are usually transient, lasting from hours to days. A pure carpal tunnel syndrome (CTS) presentation is only rarely, if ever, a presentation of HNPP. Occasionally, a brachial plexopathy may be the presenting symptom. However, HNPP is a distinct disorder from hereditary brachial plexus neuropathy.

CMT1B AND CMT1D

Phenotypes are also quite variable in other forms of CMT1. Based on initial reports, most patients with CMT1B were thought to have the "typical" CMT phenotype described for CMT1A patients, perhaps with more pronounced calf wasting. However, it is now evident that patients with MPZ mutations actually have a wide range of phenotypes ranging from the very severe (congenital hypomyelination, presenting in utero, and Dejerine-Sottas cases, presenting in infancy) or milder "CMT2"-like cases, presenting in adulthood. The type and location of the mutation on the MPZ coding region appear to determine the severity of the neuropathy, although careful genotype-phenotype correlations remain to be performed.

(continued on page 9)

* Proprioception is the awareness of posture, movement, and changes in equilibrium, and the knowledge of position, weight and resistance of objects in relation to the body.

GIFTS WERE MADE TO THE CMTA:

IN MEMORY OF:

Larry Barnes
Doris & Jim Cramer

Jane Brieff
Mindy & Myron Strober

Gwynolyn Doble
Arthur Doble

Hazel Ferguson-Antony
Wallace & Wanda Brown

Hubert Firnhaber
Ron & Carole Andresen
Leonard & Dorothy Bahr
Anne Barstow
Rev. Steve & Corinne Bielenberg
Mrs. Muriel Biermann
Milo & Mary Ann Brekke
Karl & Dianne Buchholz
Larry & Kathy Buzzard
Maynard & Norma Campbell
Doug & Phyllis Cederberg
Curt & Mona Donahue
Natalie Firnhaber
Pastor Norb & Geanie Firnhaber
Charlene Fischer
Mrs. Margaret Goeppinger

Hubert Firnhaber (cont.)
Evelyn Grayson
Alden & Lauraine Hill
Colleen Hill
Dean & Eunice Hill
Del & Naomi Karmeier
Jim & Debbie King
Joan Kroeller
Bob Langholz
Loren & Olive Lee
Dave & Colleen Marasus
Larry & Cherie Martin & family
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Rainey & Karen Mills
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Dick & Dorene Ronish
Dorinne Ross
Mrs. Billie Rupprecht
Frank & Edith Rychlick
Sharon Sandman
Gordon & Shirley Scheer
Bob & Lois Scheve
Karl & Diana Schlichting
Marvin & Sara Schrader
Ray & Katy Sievert
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Ron & Cathy Snyder
Shirley & Kenneth Solberg
Sonderen Packaging
Evelyn Thomsen
Ken & Alice Thorstad

Hubert Firnhaber (cont.)
Pat & Carrie Travica & family
Margaret Umland
Don & Margaret Wachlin
Elsie Weichbrodt
Willis & Dorothy Wurdeman

Margaret Hendrick
Dorothy Womble

Rubin Johnson
Vivian Anderson
Barb Janisch

James Lee Martin
Patty Martinez

Rebecca Sand
Rhoda & Stephen Sand

Doug Seeloff
Doris & Jim Cramer

Ken Takayama
Sheran Clark
Douglas Higashi
Leslie Nakamatsu
June Otaguro
Marian Wong

IN HONOR OF:

Mr. & Mrs. Richard Davis
Skip & Pat Davis

Adam Gelman
Leon Gelman

Pamela Kleinman
Marsha & Ray Greenberger

Phyllis Sanders
Dr. Frank Cohen

Harold Seybert
Herbert & Rosalia Estrin
Barbara & Alvin Mass
Linda & Lloyd Sneddon

Bernice & Ed Zeller
Irving & Elaine Berg
Robert & Marion Crane
Irma & Jacob Jaffe
Joel & Myra Kahn
Elaine & Eddie Lieberfarb
Mr. & Mrs. Marv Nelson
Paula & Stan Perlowitz
Rita Reisman
Barbara & Stanley Rosen
Mrs. Natalie Roth
Janet, Jeff & Zachary Zysberg

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:_____

_____

CLINICAL FEATURES

(Continued from page 7)

Missense mutations in EGR-2 also cause variable phenotypes, probably depending on the site and nature of the specific mutation. To date, most mutations have caused severe disease, classified as Dejerine-Sottas or congenital hypomyelination. More recently, however, cases have been described with milder phenotypes which do not present until adulthood.

X-LINKED CHARCOT-MARIE-TOOTH DISEASE (CMTX)

CMTX patients usually develop symptoms in late teenage years or young adulthood. Several patients we have evaluated have been varsity athletes in high school, though they were never fast runners. Wasting calf muscles is often more pronounced in CMTX than in CMT1A patients. Interestingly, despite the more than 200 different mutations described, few if any appear to have severe Dejerine-Sottas or congenital hypomyelination phenotypes. As with CMT1A patients, abnormalities are usually slowly progressive, limited to the distal legs and hands, and do not shorten a patient's lifespan. Occasional female patients have presented in adulthood with a Chronic Inflammatory Demyelinating Polyneuropathy-like neuropathy.

CMT TYPE 2

CMT2 represents up to a third of cases with autosomal dominant CMT. In most respects, the clinical phenotype of CMT2 patients is similar to that of patients with CMT1. CMT2 patients also have distal weakness, atrophy, sensory loss, and foot deformities. In general, CMT2 patients may have a wider age range of onset and disability than those with CMT1, and CMT2 patients are more likely to maintain their deep tendon reflexes. However, it is impossible to accurately distinguish CMT1 from CMT2 patients clinically without utilizing electrodiagnostic testing. Normal or mildly slow conduction velocities are hallmarks of CMT2.

Molecular genetic studies prove CMT2 to be a heterogeneous disorder, like CMT1. Five subtypes of CMT2 (2A, B, C, D, E) have been identified by linkage analysis.

CMT2A patients have typical CMT clinical presentations with sensorimotor peripheral neuropathies. CMT2B is a predominately sensory disorder and there is debate as to whether cases should be considered under pure sensory neuropathies. CMT2C is a rare disorder in which patients have paresis of vocal cords, pupillary abnormalities, and hearing loss, in addition to other characteristics of CMT2. CMT2D is a somewhat confusing disorder because some patients appear to have sensorimotor neuropathies while others have pure motor syndromes characterized as hereditary motor neuropathy type V (HMNV). CMT2E, recently discovered, has mutations in the neurofilament light gene, which is important in the axonal transport systems.

CMT TYPE 4

CMT4, the autosomal recessive inherited neuropathies, are also a heterogeneous group of disorders. CMT4 cases are rare, usually more severe than the autosomal dominantly inherited disorders, and many patients have systemic symptoms, such as cataracts and deafness. CMT4 is separable into demyelinating (4A and 4B) and axonal (4C) forms. Affected patients become symptomatic early, with an average onset of 34 months. Unlike most forms of CMT, proximal as well as distal weakness is prominent. Motor conduction velocities are severely reduced (typically 14-17 m/sec).

Finally, a novel, severe form of CMT has been designated CMT4F and defined in a large Lebanese family in which the mutations have been found in the periaxin (PRX) gene on chromosome 19. ★

CMTA Presents New Home Page

Go to *www.charcot-marie-tooth.org* and see the newly designed home page. Among the changes are increased simplicity of movement through the site, less clutter on the first page, different uses of color and the ability of the CMTA staff to make more frequent and, therefore more current, changes to the news items.

Some of the favorite items, like the discussion forum, remain unchanged.

Please let us know how you like (or don't) the new design. We welcome any suggestions you have to make the site more user friendly. ★

Ask the Doctor

Dear Doctor:

My 37-year-old husband was diagnosed with Charcot-Marie-Tooth disease (CMT) when he was about 15 years old and about 2½ years ago began wearing AFOs. The braces have helped his lower leg problems significantly, but now he is beginning to notice problems with his hands that he describes as a delayed reaction of function. He also says there is some weakness and shaking. Is there any type of brace, etc. that may be helpful to prevent these problems with his hands?

The doctor replies:

Hand involvement in Charcot-Marie-Tooth disease (CMT) usually occurs later in life and produces weakness and wasting of the hand and finger muscles that make it difficult to extend the fingers. The thumb gets weak and rotates externally, making movements such as pinching, opening jars, buttoning, and unbuttoning very difficult. A good occupational therapist can not only teach you some hand and finger exercises, but he or she can get you some tools such as button hookers and tools to hold eating utensils and other items that will help you with some of your daily activities. If indicated, the therapist, along with your physician, can also recommend some type of splint or braces. Tremor is a frequent occurrence in CMT and is aggravated by the nicotine in cigarettes and caffeine in coffee, tea, etc. There are some medications that improve tremor, including beta-blockers and anticonvulsants. You need to consult your neurologist for this.

FOOT CARE FOR HAPPY FEET!

Foot care is very important for people with CMT. Peripheral neuropathy (nerve damage in the legs and feet) and vascular disease (poor circulation) are major factors contributing to foot problems. Together, these problems make it easy to get ulcers and infections. You can decrease your risk of infections and/or ulcers when foot problems are treated quickly. To prevent these complications, you should take good care of your feet and see your health care provider right away should any foot problems arise.

How Often Should I Have My Feet Examined?

- Every day for cuts, blisters, sores, swelling, redness or sore toenails.
- Remove your shoes and socks at each doctor visit for a thorough foot exam to look for any areas of irritation or infection.

Foot Care Tips:

- Look at your feet every day to check for sores, blisters, redness, calluses, or other problems. If you cannot bend over or pull your foot up to check your feet, use a mirror. If you cannot see well, ask someone else to check your feet.
- Wash your feet in warm water every day. Check the temperature—never use hot water. Dry your feet well, especially between your toes.
- If your skin is dry, rub lotion on your feet after you wash and dry them. Do not put lotion between your toes.
- Get treatment for corns, calluses, and warts. Do not cut corns or calluses yourself. Consult your doctor before using over-the-counter (OTC) chemicals to treat these conditions. Some products can be too strong and burn your skin.
- Trim toenails straight across. File edges with an emery board or nail file. If you cannot cut your own toenails, discuss this with your health care provider.
- Always wear socks or stockings. Do not wear socks or knee-high stockings that are too tight below your knee.
- Wear shoes that fit well. Never walk barefoot, even when you are at home. Shoes should feel comfortable when you buy them. Be sure the lining is smooth and there are no objects inside your shoes.
- Take only medicines your doctor tells you to use, including OTC medications.
- Stop smoking!

A little extra attention to your feet today may help prevent foot problems and major complications in the future. ★

From Independence Blue Cross Update

Dear Doctor:

My foot doctor requested that I submit this question. Why is it that people with Charcot-Marie-Tooth disease (CMT) have problems with the heel of the foot drying out which, in turn, causes heavy calluses and cracks that develop from inside or under the skin and work their way out to the surface? What will help improve the circulation in the area of the ankle especially when it seems that a blood discoloration is occurring?

The doctor replies:

There are no associated skin changes from Charcot-Marie-Tooth disease itself. The callus formation occurs because the foot is put into a position where there is more friction in one area and the skin's reaction to that is to form a callus. A callus usually has a dry, cracked skin appearance. Sometimes a neuropathy will affect the tone of the capillaries (the tiny blood vessels) and cause reduced blood flow to the skin, causing a discoloration like what you seem to be describing. There is not a great treatment for this, either, I'm afraid, except perhaps to make sure that there is nothing else causing your feet to be discolored. Next time you see your doctor, have him/her palpate your pulses to be sure that the large arteries are not affected by something else.

Dear Doctor:

My husband had surgery to remove a large, cancerous tumor from his stomach, but there is reason to believe he must have chemotherapy and radiation now. Are you aware of any data on how Charcot-Marie-Tooth disease (CMT) patients do during chemotherapy and radiation? Are there concerns that need to be brought to the attention of the hematologist before my husband begins the chemotherapy treatment currently being set up?

The doctor replies:

This is an important issue for Charcot-Marie-Tooth disease (CMT) patients, especially those with Type 1A, the most common form. Vincristine or other vinca alkaloids are the only chemotherapy drugs clearly of very high risk and they should be avoided especially in Type 1A (demyelinating form). Other chemotherapy drugs that are of lesser, but significant risk, and could cause an additional neuropathy or worsen CMT include: cisplatin and analogs, thalidomide, suramin, taxol (paclitaxel, docetaxel), misonidazole.

Other chemotherapy drugs less commonly used of lesser or low risk are: 5-azacitidine, 5-fluouracil, cytarabine (high dose), etoposide (VP-16), gemcitabine, hexamethylmelamine, ifosfamide.

MAKE CHOICES FOR YOUR CARE:

Spell Out Your Wishes in Advance

Do you have a living will or an advance directive—a document that spells out how you want to be cared for in the event that you cannot articulate your wishes?

Many of us do not think about such issues until we are admitted to a hospital. That is when, in accordance with federal law, we are asked if we have signed any such documents.

Of course, that is probably the worst time to make such decisions, particularly if it is an emergency and you are in no shape to think clearly. Imagine what a burden it can be for your family if you have not made your wishes known ahead of a debilitating illness or injury.

It may be best to discuss these matters with your family, your physician, and your attorney ahead of time.

Advance directives can be spelled out in several ways:

- **Living Will**

This document expresses your wishes should you become terminally ill or be in a persistent vegetative state. You state whether you would want to be kept alive through such measures as tube feeding, artificial respiration, or heart resuscitation. For a living will to be valid, it must be in writing, signed, dated, and witnessed by two adults. Consult your attorney.

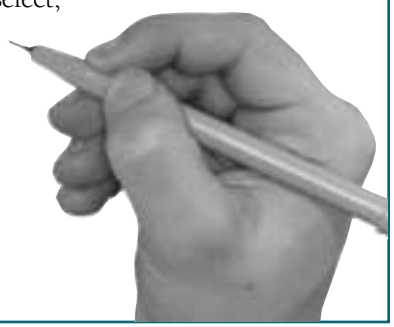
- **Health Care Agent**

This is someone you appoint to make decisions about your health care in the event that you are unable to make decisions yourself. You may specify to your agent what procedures you do or do not want, but this is not necessary. Again, contact your attorney to draft the official document needed to appoint a health care agent.

- **Durable Power of Attorney for Health Care**

This document allows you to designate a health care surrogate to make decisions for you even if you are temporarily unable to express your wishes. This can be part of a general durable power of attorney that allows your surrogate to make decisions on your behalf in virtually all matters—legal, personal, and financial. To prepare one, consult your attorney.

Whatever advance directives you select, give copies to your family, your attorney, your physician, and other health care providers, and ask that they be made part of your permanent medical record. ★



Support Group News

Pennsylvania—Johnstown

The Johnstown CMT Support Group held its bimonthly meeting in the Myron C. Williams Conference Center at the Conemaugh Health System (CHS), Saturday, September 6. Co-support leader Jeana Sweeny discussed her recent experience as the first patient to complete the new Conemaugh CMT program. She is a veteran of other CMT programs and gave Conemaugh rave reviews. She was very impressed with the staff's expertise and compassion.

Jan Goodard, from CHS, talked about the CMT program and other programs, including chronic pain and healthy living with a chronic condition.

Meghan Stahl spoke of her trip with her father to Wayne State University to see Dr. Shy and his staff. She was very impressed and enthusiastic about her experience. Interestingly, she said, the clinic included a respiratory test in the program.

The time of the meetings was debated and the group may schedule a weekday evening meeting. They also discussed the CHS Neuroscience opening, the September Awareness month activities, and the CMT Halloween Dance. The group wanted to learn more about AFOs, so they will have an expert in the field at their next meeting.

Pennsylvania—Philadelphia Area

On Saturday, September 13, 2003, the Philadelphia area support group enjoyed a presentation by Keith Marcusic and representatives from EZ Gait. They demonstrated the uses of their bracing system and then invited members of the support group to try on sneakers with the braces attached. Most of the attendees did try the braces and many were delighted and amazed with the comfort and help they received with their foot drop. The brace is not designed to prevent ankle roll-over or to help people who have no muscle left to help them walk, but for those who were less affected, it was an exciting alternative to conventional AFOs. Support group leader Amanda Young opened the meeting by having the attendees introduce themselves. Despite the terrible weather, approximately 35 people attended the meeting, some for the first time. The group's next meeting, in October, will feature CMT expert and physical therapist, Dr. Carol Oatis.

Dennis Devlin tried on and demonstrated the use of the EZ Gait bracing system. Members of the Philadelphia area support group enjoyed the chance to try the bracing for themselves.

Missouri—Kansas City

The Kansas City Area Support Group hosted an all-day seminar for those with CMT, spouses, and caregivers, on Saturday, October 11. The Kansas University Medical Center Neurology Department provided meeting space at the Landon Center for the Aging, as well as making available members of their staff to discuss areas of concern for those with CMT.

In addition to background data on CMT, subjects touched upon included genetics, MDA clinic information, blood tests, insurance billings, pedorthics, moderate bracing, children's questions, and pain and medication information. The seminar concluded with a brief discussion about the Americans with Disabilities Act.

More than 55 members and guests attended this first all-day support group gathering and voiced a desire to have another in the spring of 2004.

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: 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, 502-695-3338
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

Minnesota—Twin Cities
Place: Call for location
Meeting: Quarterly
Contact: Maureen Horton, 651-690-2709
Bill Miller, 763-560-6654
E-mail: mphorton@qwest.net, wmiller7@msn.com

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: Beverly Wurzel, 845-783-2815
E-mail: 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

Ohio—NW Ohio
Place: Medical College of Ohio
Meeting: Quarterly
Contact: Jay Budde, 419-445-2123 (evenings)
E-mail: jbudde@fm-bank.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@hotmail.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: Penn Towers Hotel Conference Room
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

WRITE TO US!

Pat Dreibelbis, Editor

The CMTA Report

CMTA

2700 Chestnut Pkwy.

Chester, PA 19013

The CMTA reserves the right to edit letters for space.

The CMTA Report is published by the Charcot-Marie-Tooth Association, a registered non-profit 501(C)(3) health organization.

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.

Photo on page 10 by Damon Hart-Davis/DHD Multimedia Gallery. Photos on pages 11 and 15 by Ian Britton, courtesy of FreeFoto.com.

Letters

Dear CMTA,

Thank you for the article "Helpful Things that Work" in the summer 2003 CMTA Report. It is a good idea for people to share ideas that make daily living easier.

A product that I have found very helpful is a coiled plastic shoelace that turns laced shoes into slip-ons. The ones I have at present are called "Curly Laces" by Ten Seconds of Hickory, NC. WalMart or Kmart sell them. They provide enough flexibility for comfort, yet are snug enough for support, without the daily struggle to tie shoelaces.

—B. L., Canton, IL

■

Dear CMTA,

Just a note to tell you that I filled out the questionnaire on physical therapy noted in your last newsletter.

While the issue of exercise and CMT is one I'm struggling with, I felt the questionnaire failed to ask a key question: what other diseases and health problems do you have? I have CLL (chronic lymphocytic leukemia) and I never know, nor do my doctors, how much of my fatigue is due to which condition.

One should also note that while one may "not feel better" after accupuncture or massage, which I find helpful, the issue can be to prevent "feeling worse," which can be a plus with exercise, as well.

But, thanks for encouraging research.

—I. J., Bronx, NY

■

Dear CMTA,

I found this printed in a church bulletin and thought it was something the membership might enjoy.

"A farmer had some puppies he needed to sell. He painted a sign advertising the new pups. Then he set about nailing it to a post on the edge of his yard. As he was driving the last nail to the post, he felt a tug on his overalls. He looked down into the eyes of a little boy.

"Mister," he said. "I want to buy one of your puppies."

"Well," said the farmer, as he rubbed the sweat off the back of his neck, "these puppies come from fine parents and cost a good deal of money."

The boy dropped his head for a moment. Then, reaching deep into his pocket, he pulled out a handful of change and held it up to the farmer.

"I've got thirty-nine cents. Is that enough to take a look?"

"Sure," said the farmer. With that, he let out a whistle. "Here, Dolly!" he called out. Out from the doghouse and down the ramp ran Dolly followed by four little balls of fur.

The little boy pressed his face against the chain-link fence. His eyes danced with delight. As the dogs made their way to the fence, the little boy noticed something else stirring inside the doghouse. Slowly, another little ball of fur appeared, this one noticeably smaller. Down the ramp it slid. Then, in a somewhat awkward manner, the little pup began hobbling toward the others, doing his best to catch up.

"I want that one," the little boy said, pointing to the runt. The farmer knelt down at the boy's side and said, "Son, you don't want that puppy. He will never be able to run and play with you like these other dogs would."

With that, the little boy stepped back from the fence, reached down and began rolling up one leg of his trousers. In doing so, he revealed a steel brace running down both sides of his leg, attaching itself to a specially made shoe. Looking back at the farmer, he said, "You see, sir, I don't run too well myself and he will need someone who understands."

■

Dear CMTA,

I opened my October *Scientific American* last night and saw on page 37 a full-color, full-page eye-catching ad on CMT awareness. WOW! The magazine is the oldest science magazine in the U.S., with a circulation of 600,000 and a readership of 4,000,000. Allison Moore, through her Hereditary Neuropathy Foundation, placed the ad as part of her campaign to promote September as CMT Awareness Month and raise CMT awareness. She is also distributing posters and submitting articles and public service announcements to newspapers and other media outlets. You can thank her at allison@hereditaryneuropathy.org or visit her website at http://www.hereditaryneuropathy.org/

—J. G., Johnstown, PA

■

Air Travel for Persons with Disabilities

(This information was supplied by Jean Ryan, who thought it would be a good follow-up to a previous article on the difficulties people encounter when traveling. Jean writes: "I have an implanted medical device and I travel a lot, but I've never had any trouble with security over it, possibly because I travel in a wheelchair. Maybe it's a matter of being lucky. In the last year, I've been to Turkey, California, Minnesota, and Costa Rica and since I live in New York, I go through a lot of security checks everywhere. I bring my passport for ID no matter where I'm flying. No longer are guide dogs allowed to be separated from their owners.")

A Toll-Free Hotline for disabled travelers has been in operation since August 2003 and is available for callers from 7 a.m. to 11 p.m. Eastern time, seven days a week. It is currently not being fully utilized. The Hotline serves two main purposes: 1) education and 2) assistance in resolving disability-related air travel problems.

Many disabled air travelers are not aware of their rights and the Hotline, in part, exists as an educational service to inform air travelers with disabilities about their rights under the Air Carrier Access Act and the Department's implementing regulations 14 CFR Part 382 (Part 382). Hotline operators are well versed in the ACAA and Part 382 and can provide callers with on-the-spot general information about the rights of air travelers with disabilities. The Hotline operators also respond to requests for printed consumer information about air travel rights of the disabled.

The Hotline can also assist air travelers with disabilities in resolving real time or upcoming issues with air carriers. The purpose of "real-time" assistance is to facilitate airline compliance with DOT's rules by suggesting to the passenger and the airline involved alternative customer service solutions to the problem. The airline remains responsible for deciding what action will be taken to resolve the issue in accordance with the ACAA and Part 382. Generally, if a caller has a real time problem or an upcoming issue with an air carrier, a Hotline Duty Officer will contact that air carrier and attempt to resolve the issue. For example, there have been a number of incidents in which Hotline Duty Officers have contacted air carriers and convinced them to accept service animals and electric wheelchairs on board flights, to stow folding wheelchairs in the cabin, and to provide requested wheelchair assistance.

Air travelers who want information about the rights of persons with disabilities in air travel or who experience disability-related air travel service problems may call the Hotline to obtain assistance at: 1-800-778-4838 (voice) or 1-800-455-9880 (TTY).

Air travelers who want DOT to investigate a complaint about a disability-related issue still must submit their complaint in writing via e-mail at airconsumer@ost.dot.gov or postal mail to:

Aviation Consumer Protection Division
U.S. Department of Transportation
400 7th Street, S.W.
Washington DC 20590 ★

PRODUCTS THAT WORK

I just bought a pair of Foot-Up Dynamic Ankle-Foot orthoses (available from www.isports.com) and they are wonderful. I avoided wearing my AFOs but these are great. They are lightweight and easy to put on and take off. I think they are a wonderful alternative to plastic AFOs. *—D. S., Seabeck, WA*

I use a carpenter's apron to carry small items up or down the stairs in my home. The many pockets leave my hands free to hold on to the railing and allow me to make one trip to do spot cleaning and other chores. The aprons can be bought at Home Depot or other similar stores. (Apron at left is from coverallstore.com.)

—E. R., Media, PA

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

CMTA

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