

SUMMER 2000

The CMTA Report

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

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The Moore Family Bequeaths \$99,471.05 to CMTA

The estate of James Thomas Moore of California has provided the Charcot-Marie-Tooth Association with the largest gift the organization has received from one individual in its 17 years of existence—\$99,471.05.

James was born in Kansas City, Missouri, on January 10, 1923. He served in the Army Air Force working as a mechanic during World War II. After the war, he worked at the North Island Naval Air Station as a civilian employee, working as a supervisor of mechanics until his retirement. James died on December 30, 1999.

His decision to donate his estate to the Charcot-Marie-Tooth Association was based on the fact that his mother, his sister, Marie Stout, of Ohio, and he were all diagnosed with CMT. He discussed his estate planning with his sister, who urged him, since he had no children, to donate to the CMTA so that research might help in finding a cure for CMT.

In addition to funding research, both Marie and James' brother, W.C. Moore, who was his executor, felt that a portion of the gift should be designated to help with the repairs to the office and equipment resulting from the flooding from Hurricane Floyd. For that reason, the gift was divided into two parts: \$74,603.29 for research and \$24,867.76 for operations.

In honor of this extremely generous and selfless gift to the CMTA, the association plans to recognize Mr. Moore by naming one of this year's post-doctoral research grants. The CMTA's James Thomas Moore Post-Doctoral Fellowship.



The late James Thomas Moore of California has bequeathed the largest single gift to the CMTA in its history.

Remembering the CMTA in your will helps the organization to achieve its goal of finding a cure for CMT and of improving the lives of those currently living with the disorder. If you have questions about making a bequest to the CMTA, please call the office for further information.

INSIDE: A bibliography of recent publications by the CMTA Medical Advisory Board Members.

■ UPCOMING CMT PATIENT/FAMILY CONFERENCE IN PENNSYLVANIA

A CMT patient/family conference is scheduled at the University of Pennsylvania on October 7, 2000. Dr. Steven Scherer, of the CMTA's Medical Advisory Board, is the organizer and more details will be available within a month. The conference will include discussions on the neurology of CMT, surgeries, therapies and other practical topics. If you do not receive an application in the mail in September and are interested in attending, please contact the CMTA office.



ADDITIONAL GIFTS WERE MADE TO THE CMTA...

IN MEMORY OF:

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Jerry & Debra Bank

Lois Basquill

Mr. & Mrs. Thomas Alberino
George, Fred & Jody Baker
Jay & Mary Beebe
Joyce Camp
Gloria Capeilli
Romona Cortese
Barbara & Ted Dumonski
Frances Ferguson, R.N. & 8-8 Staff
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Dubby & Bernie Bernstein's 60th Anniversary

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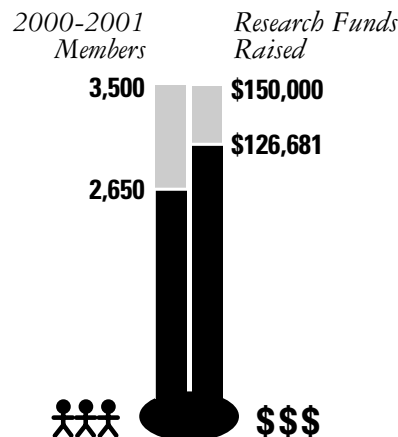
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Leah Vaknin

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Our Research Fundraising Progress as of July 15, 2000:



In addition to the Moore family gift, a very generous, combined gift of \$20,000 from Dr. and Mrs. Robert Buch has significantly increased the research funds' total.



SPECIAL OFFER: Get All The Facts...

For the first time, you can get the entire CMT Facts Series (I, II, III, and IV) for the low price of \$16.00, plus \$3.00 for shipping and handling (see the order form at right). **This is a special offer being made to "active" dues-paying members of the CMTA.**

Purchased separately, the CMT Facts Series would cost \$21.00 for active members—that's a \$5.00 savings for the whole set. Please note, only Facts I and II are available in Spanish.

Do you already have one or two issues in the series? Why not buy the full set and share those you already have with family members? You might consider giving a set to your doctors or making the series available to a local clinic or library to help raise awareness about CMT disorders.

The newest in the series, *Facts IV*, is 32 pages in length with six different subject areas including: Medical Information and Research, CMT Survey Results, Living with CMT, Children and CMT, Legal Information, and Q&A.



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If you are unsure as to whether you are current with your member dues, please call the office at 1-800-606-CMTA.

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Doctors and Patients ... Working

By virtue of an email, your editor became aware of a novel and exciting research project that joins a large family with a possible CMT1/CMT2 overlap syndrome with a member of our Medical Advisory Board. The Meier/Haislip/Minzer family and Dr. Florian Thomas are embarking on a very promising collaboration to describe the phenotypes and genotypes of this unique overlap syndrome.

We were first contacted by Lisa Minzer, who was diagnosed with CMT2 in 1995. All six of her siblings and her mother, Carol Haislip, founder of the CMT support group in St. Louis, MO, have CMT. The symptoms of CMT within her family range from moderate to severe. The size of the family caught the attention of Dr. Florian Thomas of St. Louis University and, along with some of his colleagues at SLU, he has embarked on a large-scale, long-term study of Lisa's immediate and extended family members. Curiously, Lisa has two sets of male twin siblings, all of whom are severely affected, much more so than the females, and four generations of family members with CMT.

Carole Haislip first met with Dr. Thomas in the spring of 1997 when she was attempting to form a support group for the St. Louis area. Dr. Thomas was instrumental in getting the group started, including providing a place for the group to meet and arranging for guest speakers. In the following months, he saw three of her sons as patients in his office. In October of 1998,

Dr. Thomas attended the Third International Conference on CMT in Montreal, Canada, and presented some of his electromyography and nerve conduction study results to other researchers. Several of them agreed that the family had unusual features, including characteristics of both CMT1 and CMT2. The large size of the family made the search for the causative gene seem very promising.

The purpose of Dr. Thomas' study is to determine the genetic marker for CMT2. This has already been discovered for several forms of CMT1. Since CMT2 affects a smaller number of individuals, it was a stroke of luck for him to happen upon such a large group to study. Because the family is so large, however, it was difficult to get everyone to show up for blood work and nerve conduction studies, so he decided to go to them.

On Easter Sunday, Dr. Thomas and two of his fellow neurologists visited Carol Haislip's home, set up an examining room and blood drawing area and did neurological exams and took blood samples from all the family members. It was, according to Lisa, "quite a surreal experience to have all these doctors and medical equipment underfoot just after the annual Easter egg hunt." The living room and bedroom were converted into examination rooms and the kitchen became a blood-drawing lab. A lot was accomplished. Nearly everyone present underwent a clinical neurological exam and many had blood drawn for the study.

On Mother's Day weekend, Dr. Thomas and three other doctors flew to tiny Menasha, Wisconsin (Carol Haislip's hometown) and did similar tests on her parents, siblings, nieces, nephews and other extended family members. The CMT in the family is believed to have originated with Genevieve Meier, who died 75 years ago at age 24. To date, some 65 people have been clinically examined in Dr. Thomas' study, most of them direct descendants of Genevieve Meier.

Of those 65, 36 completed a standardized neuropathy questionnaire, 40 underwent nerve conduction studies and electromyography, 1 had a lumbar puncture, 2 had sural nerve biopsies, and 50 had blood drawn for DNA testing. CMT was diagnosed in 21 subjects, including 6 children of ages 5, 7, 8, 9, 10 and 14 years.

On June 15, 2000, Dr. Thomas gave his first presentation of his initial research findings at St. Louis University Medical School, where he is an Associate Professor of Neurology. Even at this

Drs. Thomas and Kinsella pose among just some of the many Meier family members who volunteered for their study.



Together on Research

stage of the study, he has found information that was previously unknown or misunderstood about this condition, including age of onset. Dr. Thomas hopes to extend the study for another ten years in order to observe the progression of the disease in the affected individuals.

The magnitude of the Meier family's commitment to this research project is inspiring and Dr. Florian Thomas offered some points to consider with regard to being part of a major research study:

"First, it is important for patients to see what they can do to further research. But it is work for them, too. Families must be precise when they gather family-tree information.

"They must also be patient with the doctors. You can not imagine how many phone calls I made to Lisa and her family. I must have gotten on their nerves, but they were kind enough not to show it.

"They must make themselves available. I ruined Easter Sunday and Mother's Day for this family.

"They must consider if they really want to go ahead with somewhat uncomfortable tests such as electromyography and nerve conduction velocity tests, and definitely uncomfortable tests such as biopsies and spinal taps.

"They must think about whether they want to subject their children to tests and whether they want to accept the small risk of having



inherited conditions listed on their children's records.

"I cannot be exuberant enough about this family's commitment. It is a testimony to the good relationships among themselves. To have their grandparents get into a bus so that I can do a biopsy on them is beyond all expectations.

"Other patients might be disappointed that no researcher wants to do such a study on them. This is probably because they already have a molecular diagnosis or because their families are too small for linkage studies."

Once this study is complete and Dr. Thomas publishes his findings, we will print a follow-up article in the newsletter.

Dr. Florian Thomas, Dr. Yi Pan, Dr. Francisco Gondim, Carole Haislip and Dr. Laurence Kinsella relax after doing blood samples and nerve biopsies.

CALL FOR PARTICIPANTS

NIH Study: Familial Vocal Fold Paralysis

Editor's note: A representative of NIH called the office to request this study be listed in our newsletter because of the incidence of vocal cord paralysis in CMT patients with Type 2C.

Researchers at the National Institute of Neurological Disorders and Stroke, NIH, seek affected and unaffected family members, ages 5 to 75, with vocal fold paralysis for a research study to improve understanding of the disorder's pathophysiology and to relate it to the genetic pattern of inheritance for this rare, potentially life-threatening, disorder. The research study takes place at the NIH Clinical Center in Bethesda, MD and is study number 96-N-0089. The study is carried out under testing and safety standards of the US Department of Health and Human Services.

NIH provides study-related tests, examinations, counseling, and travel and lodging expenses at no cost to participants. Families may qualify if the condition is due to an unknown cause and has occurred over at least two generations. Families may not qualify if the condition was caused by traumatic injury.

Referring physicians receive test information and patients return to their physician's care at study completion. For more information or to speak to the investigator, please call 1-800-411-1222 or email: prpl@cc.nih.gov

A Bibliography of Recent Publications By Members of the Medical Advisory Board

Editor's note: The following publications were found in PubMed, a service of the National Library of Congress, and represent only the past two years of published work by members of the CMTA's Medical Advisory Board and our post-doctoral grant recipients.

- Bird TD.** "Historical perspective of defining Charcot-Marie-Tooth type 1B." *Annals of the New York Academy of Science*. 1999 Sep 14; 883:6-13.
- Boerkoel CF, Inoue K, Reiter LT, Warner LE, Lupski JR.** "Molecular mechanisms for CMT1A duplication and HNPP deletion." *Annals of the New York Academy of Science*. 1999 Sep 14; 883:22-35.
- Butefisch C, Gutmann L, Gutmann L.** "Compression of spinal cord and cauda equina in Charcot-Marie-Tooth disease type 1A." *Neurology*. 1999 Mar 10; 52(4):890-1.
- Chance PF.** "Overview of hereditary neuropathy with liability to pressure palsies." *Annals of the New York Academy of Science*. 1999 Sep 14; 883:14-21.
- Chance PF.** "Molecular genetics of hereditary neuropathies." *Journal of Child Neurology*. 1999 Jan; 14(1):43-52.
- Chance PF.** "Survey of inherited peripheral nerve diseases." *Electroencephalography and Clinical Neurophysiology Supplement*. 1999; 50:121-8.
- De Jonghe P, Nelis E, Timmerman V, Lofgren A, Martin JJ, Van Broeckhoven C.** "Molecular diagnostic testing in Charcot-Marie-Tooth disease and related disorders." Approaches and results. *Annals of the New York Academy of Science*. 1999 Sep 14; 883:89-96.
- De Jonghe P, Timmerman V, Ceuterick C, Nelis E, De Vriendt E, Lofgren A, Vercruyssen A, Verellen C, Van Maldergem L, Martin JJ, Van Broeckhoven C.** "The Thr124Met mutation in the peripheral myelin protein zero (MPZ) gene is associated with a clinically distinct Charcot-Marie-Tooth phenotype." *Brain*. 1999 Feb; 122 (Pt 2): 281-90.
- De Jonghe P, Timmerman V, Nelis E, De Vriendt E, Lofgren A, Ceuterick C, Martin JJ, Van Broeckhoven C.** "A novel type of hereditary motor and sensory neuropathy characterized by a mild phenotype." *Archives of Neurology*. 1999 Oct; 56(10): 1283-8.
- Ellsworth RE, Ionasescu V, Searby C, Sheffield VC, Braden VV, Kucaba TA, McPherson JD, Marra MA, Green ED.** "The CMT2D locus: refined genetic position and construction of a bacterial clone-based physical map." *Genome Research*. 1999 Jun; 9(6):568-74.
- Fischbeck KH, Abel A, Lin GS, Scherer SS.** "X-linked Charcot-Marie-Tooth disease and connexin32." *Annals of the New York Academy of Science*. 1999 Sep 14; 883:36-41.
- Garcia CA.** "A clinical review of Charcot-Marie-Tooth." *Annals of the New York Academy of Science*. 1999 Sep 14; 883:69-76.
- Griffin JW, Sheikh K.** "Schwann cell-axon interactions in Charcot-Marie-Tooth disease." *Annals of the New York Academy of Science*. 1999 Sep 14; 883:77-90.
- Hahn AF, Bolton CF, White CM, Brown WF, Tuuha SE, Tan CC, Ainsworth PJ.** "Genotype/phenotype correlations in X-linked dominant Charcot-Marie-Tooth disease." *Annals of the New York Academy of Science*. 1999 Sep 14; 883:366-82.
- Hwang MY, Kang YJ, Kim YH, Scherer SW, Tsui LC, Sohn U.** "Generation and chromosome mapping of expressed sequence tags (ESTs) from a human infant thymus." *Genome*. 1999 Jun; 42(3):457-64.
- Inoue K, Tanabe Y, Lupski JR.** "Myelin deficiencies in both the central and the peripheral nervous systems associated with a SOX10 mutation." *Annals of Neurology*. 1999 Sep; 46(3):313-8.
- Irobi J, Timmerman V, De Jonghe P, De Vriendt E, Van Broeckhoven C, Beuten J.** "Construction of a PAC contig within the distal hereditary motor neuropathy type II candidate region at 12q24." *Annals of the New York Academy of Science*. 1999 Sep 14; 883:463-5.
- Jetten AM, Suter U.** "The peripheral myelin protein 22 and epithelial membrane protein family." *Progress in Nucleic Acid Research and Molecular Biology*. 2000; 64:97-129.
- Kamholz J, Awatramani R, Menichella D, Jiang H, Xu W, Shy M.** "Regulation of myelin-specific gene expression. Relevance to CMT1." *Annals of the New York Academy of Science*. 1999 Sep 14; 883:91-108.
- Kashork CD, Chen KS, Lupski JR, Shaffer LG.** "Prenatal diagnosis of Charcot-Marie-Tooth disease type 1A." *Annals of the New York Academy of Science*. 1999 Sep 14; 883:457-9.
- Kashork CD, Lupski JR, Shaffer LG.** "Prenatal diagnosis of Charcot-Marie-Tooth disease type 1A by interphase fluorescence in situ hybridization." *Prenatal Diagnosis*. 1999 May; 19(5):446-9.
- Keller MP, Chance PF.** "Inherited neuropathies: from gene to disease." *Brain Pathology*. 1999 Apr; 9(2):327-41.
- Keller MP, Chance PF.** "Inherited peripheral neuropathy." *Seminars in Neurology*. 1999; 19(4):353-62.
- Keller MP, Seifried BA, Chance PF.** "Molecular evolution of the CMT1A-REP region: a human- and chimpanzee-specific repeat." *Molecular Biology and Evolution*. 1999 Aug; 16(8):1019-26.
- Kochanski A, Timmerman V, Jedrzejska H, Ryniewicz B, Lofgren A, De Vriendt E, Van Broeckhoven C, Latos-Bielenska A, Hausmanowa-Petrusewicz I.** "Mutation screening of Charcot-Marie-Tooth patients in Poland." *Annals of the New York Academy of Science*. 1999 Sep 14; 883:493-6.
- Krajewski K, Turansky C, Lewis R, Garbern J, Hinderer S, Kamholz J, Shy ME.** "Correlation between weakness and axonal loss in patients with CMT1A." *Annals of the New York Academy of Science*. 1999 Sep 14; 883:490-2.
- Kumar N, Cole J, Parry GJ.** "Variability of presentation in hereditary neuropathy with liability to pressure palsy results in underrecognition." *Annals of the New York Academy of Science*. 1999 Sep 14; 883:344-50.

on Charcot-Marie-Tooth Disease Award and Grant Recipients (1999-2000)

- Lewis RA, Shy ME.** "Electrodiagnostic findings in CMTX: a disorder of the Schwann cell and peripheral nerve myelin." *Annals of the New York Academy of Science*. 1999 Sep 14; 883:504-7.
- Lewis RA, Sumner AJ.** "Electrophysiologic features of inherited demyelinating neuropathies: a reappraisal." *Annals of the New York Academy of Science*. 1999 Sep 14; 883:321-35.
- Lin GS, Glass JD, Shumas S, Scherer SS, Fischbeck KH.** "A unique mutation in connexin32 associated with severe, early onset CMTX in a heterozygous female." *Annals of the New York Academy of Science*. 1999 Sep 14; 883:481-4.
- Lofgren A, De Vos A, Sermon K, Liebaers I, Van Steirteghem A, Van Broeckhoven C.** "Preimplantation diagnosis for Charcot-Marie-Tooth type 1A." *Annals of the New York Academy of Science*. 1999 Sep 14; 883:460-2.
- Lupski JR.** "Recessive Charcot-Marie-tooth disease." *Annals of Neurology*. 2000 Jan; 47(1):6-8.
- Lupski JR.** "Charcot-Marie-Tooth polyneuropathy: duplication, gene dosage, and genetic heterogeneity." *Pediatric Research*. 1999 Feb; 45(2):159-65.
- Mackin GA, Gordon MJ, Neville HE, Ringel SP.** "Restoring hand function in patients with severe polyneuropathy: the role of electromyography before tendon transfer surgery." *Journal of Hand Surgery [Am]*. 1999 Jul; 24(4):732-42.
- Marques W Jr, Hanna MG, Marques SR, Sweeney MG, Thomas PK, Wood NW.** "Phenotypic variation of a new P0 mutation in genetically identical twins." *Journal of Neurology*. 1999 Jul; 246(7):596-9.
- Menichella DM, Xu W, Jiang H, Sohi J, Vallat JM, Baron P, Kamholz J, Shy M.** "The absence of myelin P0 protein produces a novel molecular phenotype in Schwann cells." *Annals of the New York Academy of Science*. 1999 Sep 14; 883:281-93.
- Mersyanova IV, Ismailov SM, Polyakov AV, Dadali EL, Fedotov VP, Nelis E, Lofgren A, Timmerman V, van Broeckhoven C, Evgrafov OV.** "Screening for mutations in the peripheral myelin genes PMP22, MPZ and Cx32 (GJB1) in Russian Charcot-Marie-Tooth neuropathy patients." *Human Mutation*. 2000; 15(4):340-7.

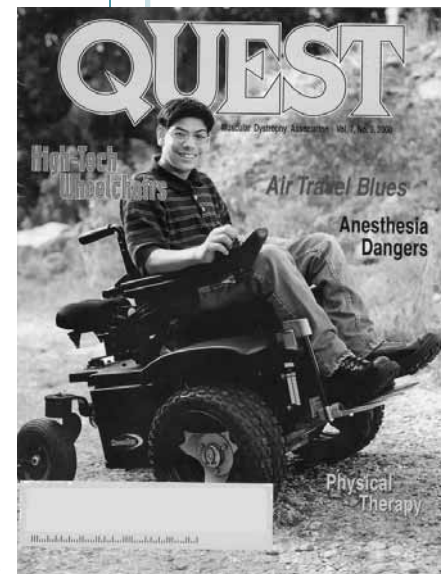
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MDA's *Quest* Provides Excellent CMT Information

Check out the most recent issue of *Quest*, the publication of the Muscular Dystrophy Association, for an article called, "Coping with Anesthesia." If you are considering any kind of surgery, from major to dental, this article is a "must-read" because it discusses problems such as malignant hyperthermia (seriously high temperature), which is rare, but can be more common with some of the muscular dystrophies; potassium release in dangerous amounts from using succinylcholine, especially in ALS and SMA patients; and the use of local anesthesia and regional anesthesia, which directly affect nerves, for patients with CMT and Dejerine Sottas disease. The article can be found on page 32 of volume 7, number 3 or online at www.mdaua.org in the section on *Quest*, the current issue.

Another excellent article in *Quest*, Volume 7, number 3, discusses the issue of loss and grief as it affects someone with a neuromuscular disease. The title, "Where there's loss, there's grief" summarizes the author's theme that even when people don't recognize the feelings they have as grief, the loss of abilities does deserve to be treated as legitimate grief. In a section entitled, "Let Yourself Feel Your Emotions, Whatever They Are," a Charcot-Marie-Tooth patient, Bill Quesinberry of Denver, Colorado, talks about getting tired of dealing with people's stares and questions. For a time, he avoided social situations by just staying home. But, his withdrawal was brief. During the time of protecting himself from others, he reflected on his situation and developed a new attitude. As he said, "I just decided that I have a great life now. I live in a gorgeous place with gorgeous views. I go to the theater, we go out to dinner. I love the volunteer work I do. Quesinberry says, "I just decided, I never cared what they thought before. Why should I care now? I realize that people can react to me, and that's fine." Another CMT patient is featured in the article and the techniques that the author suggests are valid ones for anyone dealing with loss.

If you do not receive the magazine *Quest* you can request a subscription from the MDA at 1-800-572-1717. It is free for registered clients of MDA's clinics or it is available for \$12 to anyone who would like to receive it bimonthly.



Special Education Evaluation...

Complying with IDEA (Individuals with Disabilities Education Act)

Editor's note: This information comes from the PACER Center and the FAPE project (Families and Advocates Partnership for Education), Minneapolis, MN.

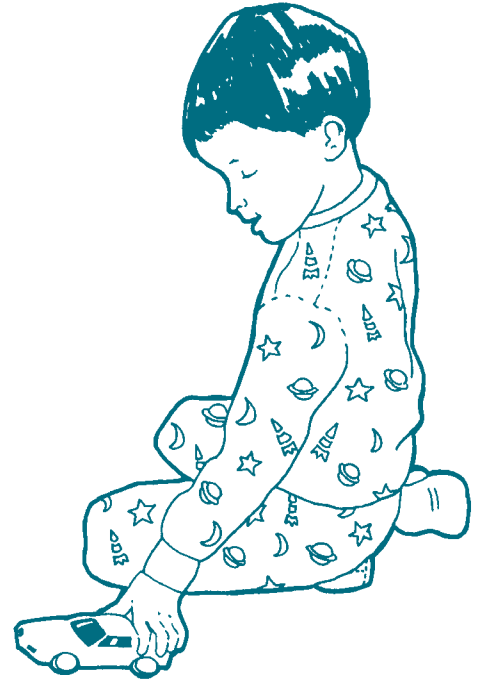
Before your child receives special educational services, the Individuals with Disabilities Education Act (IDEA) requires that he or she have an evaluation. An evaluation includes information from parents, any special medical concerns, interviews with parents and school staff, and specific tests. The results of the evaluation will be used to determine if your child has a disability and needs special education and related services help in school. You must give your consent in writing before the first evaluation can take place.

The evaluation:

- must be done by trained and knowledgeable persons;
- must be in all areas related to the suspected disability;
- is more than just one test or assessment procedure;
- must be in your child's native language unless clearly not possible;
- must not discriminate against your child.

Some questions that parents can ask the school staff about evaluation are:

1. Why do you want to evaluate my child?
2. What do you think you may find from the evaluation?
3. What kinds of tests will you do?
4. In what areas will my child be tested?
5. Will the tests you use discriminate against my child based on Race? Disability? Culture?
6. How do you know that the tests do not discriminate?
7. What will happen if my child is not evaluated? Will he or she still get help for the problems you have identified?
8. By what date will you give me a written copy of the evaluation results?
9. What steps should I take if I do not agree with the evaluation results?



If you disagree with the school district's evaluation results, you have a right to:

- have someone outside the school district evaluate your child (this is called an Independent Educational Evaluation [IEE]);
- help select the person or persons who will do the testing;
- have the testing done at no cost to you or your family. (The school district must either have your child evaluated at no cost to you, or show, at a hearing, that its evaluation is appropriate.)

If your child already receives special education, he or she must have a re-evaluation at least every three years or more often if needed. The purpose of the re-evaluation is to:

- see if your child still has a disability and needs special education or related services;
- identify how your child is doing in school and identify any educational needs;
- determine if any changes need to be made in the child's IEP (Individual Education Program) to help your child to meet the annual goals and objectives in the IEP and to participate, as appropriate, in the general curriculum.

Chronic Pain

The following information was taken from the Internet source, *Information about Chronic Pain* located at <http://209.41.17.84/info/paininfo.html>

WHAT IS PAIN?

Pain is an unpleasant sensory and emotional experience associated with actual or potential tissue damage or described in terms of such damage. Pain is suffering and it is familiar to everyone and yet it is so complex and subjective it cannot easily be described or treated.

HOW IS PAIN CLASSIFIED?

There are generally two different types of pain: acute and chronic. Acute pain is described as temporary and frequently occurs as a result of an injury to the body. Usually, acute pain disappears when the injury is healed or the cause of the pain is eliminated as in postoperative pain or labor contractions. Chronic pain may be divided into three classes: 1) chronic non-malignant pain such as low back pain and rheumatoid arthritis; 2) chronic intermittent pain such as migraine headaches; and 3) chronic malignant pain such as the pain associated with cancer and AIDS. Chronic pain causes severe physical as well as psychological suffering.

PAIN—A NEW FIELD OF MEDICINE

The study of pain, or algology, is a new field. In the past, healthcare professionals have not been educated on the proper identification, assessment and management of pain. In general, there has been a lack of understanding about the pain process, concerns about respiratory depression and addiction and incomplete knowledge regarding drug pharmacology and the mechanisms of other treatment methods. Too often, easing the hurt took a back seat to seeking an elusive cure and, as a result, a patient's pain often went unrecognized and untreated. Physicians still have few weapons against pain, and each has drawbacks. Fortunately, a number of new alternatives are on the horizon and physicians believe that these new drugs will be safer and more effective than current therapies.

PATIENT GUIDELINES FOR DEALING WITH PAIN

Critical to successful pain management is an accurate assessment of the pain and involvement

of the patient. Here are some guidelines for the patient:

- Talk to your doctor and nurses about your pain. Tell them how much pain you feel, where you feel it and what it feels like. Remember, pain is what you say it is; there are no right or wrong answers.
- Work with your doctor and nurses to manage your pain. Let them know what makes the pain feel better or worse and how well your treatment is working. Be sure to tell them if your pain changes over time.
- Talk to your doctor or nurses about your concerns and fears about any medications you are trying. They can help you understand your pain treatment.
- Take your medication regularly as your doctor prescribes. This will help keep your pain under control. Don't skip a scheduled dose. Ask your doctor how and when to take extra medication if your pain becomes worse.
- If you experience side effects, report these to your doctor at once. Often he or she can prescribe medications to counteract those problems, or change your medication.

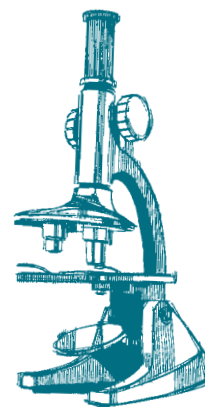
CALL FOR PARTICIPANTS

CMT and Trigeminal Neuralgia Study

Dr. James Lewis, a neurologist in private practice in rural western North Carolina, is looking for families with a rare combination of Charcot-Marie-Tooth disorder with trigeminal neuralgia. (Trigeminal neuralgia is a disorder of the fifth cranial nerve in which episodes of severe, stabbing pain affect the cheeks, lips, gums or chin on one side of the face. The pain is very brief, but so intense that the person is unable to do anything for the duration of the episode. The pain often causes wincing and twitching and is commonly called tic douloureux (literally, painful twitch.)

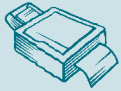
If your family has this combination of problems, please contact Dr. Lewis at 828-369-4268, fax:828-369-4488 or email:treehaus@dnet.net

Dr. Lewis hopes to have some interesting information about a specific gene defect associated with this variant situation in the near future.



THANK YOU!

Dan Charny, CMTA Board member, donated a new Hewlett Packard computer and Lexmark color printer to the office to help with our continuing restructuring following the flood.



The Trager Approach

(Editor's note: If you received the Fall 1999 newsletter, you will remember a letter from a woman in California who remarked about Trager therapy and how it helped her move her toes after a year and a half of work. Numerous members called and wanted more information, and it is now available. We present a brief overview below.)

The Trager Institute is located in Mill Valley, California, and is a non-profit, public benefit, educational corporation that represents and supports Trager psychophysical integration and Mentastics movement education. This movement therapy was developed by Milton Trager, M.D., and is usually called The Trager Approach.

There are two aspects of The Trager Approach; one in which the client is passive, referred to as the tablework, and one in which the client is active, called Mentastics. Utilizing gentle, non-intrusive, natural movements, the procedure is intended to release deep-seated physical and mental patterns and facilitates deep relaxation, increased physical mobility, and mental clarity.

A session usually lasts from 60 to 90 minutes. During the tablework session, the client is passive and lying on a comfortably padded table. The practitioner moves the client in ways he/she naturally moves, and with a quality of touch that makes the client feel he/she is mov-



ing effortlessly and freely on his/her own. Movements are never forced, so there is no pain or discomfort. The effortless movement is reinforced by the phase called Mentastics. These are simple, active, self-induced movements that the client can do on his/her own, during daily activities. They have the same intent of releasing deep-seated patterns that create stress and tension.

The Trager Institute maintains a database of certified practitioners worldwide. If you have questions or are interested in finding a therapist near you, you can call the Institute at 415-388-2688 or go to their home page at www.trager.com



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: _____

CMT Forum Planned in Boston for Young Researchers

A Charcot-Marie-Tooth research forum and symposium will be held in Boston, MA, on October 15, 2000, in conjunction with the 125th meeting of the American Neurological Association. The morning session will feature young investigators presenting their work on the Charcot-Marie-Tooth group of genetic neuropathies. Presentations will be limited to 15 minutes, with 2 or 3 minutes allotted to questions and discussion. In addition to oral presentations, some studies will be represented by poster presentations. The morning session will run from 8:30 AM to 12:00 noon. This is the



The Boston ANA meeting will provide a unique opportunity for young CMT investigators to meet and share research findings.

first time the Charcot-Marie-Tooth Association's Medical Advisory Board has sponsored such a forum for young researchers.

The afternoon session will run from 1:30 PM to 5:30 PM and will summarize the current status of genetic and clinical knowledge about CMT. Guest speakers will include Drs. Michael Shy, P.K. Thomas, Steven Scherer, Richard Lewis, James Kamholz and Angelica Hahn. An innovative use of molecular genetic engineering will be discussed, using immunogenetic models. There will be adequate time for discussions and, hopefully, new collaborations will be forged.

Cramps, Fatigue and "Twitching" Aspects of CMT

The information for this article was provided by the MDA (Muscular Dystrophy Association).

All neuromuscular diseases cause a variety of symptoms other than simple muscle weakness. Some people experience cramping; some deal with severe fatigue and still others notice their skin crawling or twitching.

A true cramp is a condition in which muscles undergo painful involuntary contractions. The classic muscle cramp is neural in origin, meaning abnormal nerve activity causes the problem rather than abnormal muscle activity. Usually such cramps occur suddenly and can often be corrected by stretching the muscle.



Fatigue is often a subjective feeling of tiredness, but it can also be an objective measurement of decline in muscle force with use. It is not to be confused with weakness. It is a feature of most neuromuscular disorders since muscles are weakened and greater energy must be expended to move them.

Fasciculation is the technical term for a twitch. Like cramps, fasciculations are caused by abnormal nerve activity, but they involve only a small portion of the muscle and they are not usually painful. When a fasciculation occurs, you often see a small muscle "jump" under the skin. All people get them now and then, especially around the eye, but fasciculations can be worsened by stress, lack of sleep and consumption of caffeine.



VISIT OUR HOMEPAGE
at

www.charcot-marie-tooth.org

The site was provided
through the generosity of



HNPP... My Journey to Recognition

By MAUREEN HORTON, St. Paul, Minnesota

When my father and I were first diagnosed with Hereditary Neuropathy with Liability to Pressure Palsies (HNPP) in 1985, we were told that there were 30 families worldwide with this disease. At the time, the chance of finding out anything about this disease and what to expect was virtually nil. So, we went on with our lives.

In truth, part of me was fascinated by this rare disease, and I tried to learn about how it would affect me. Another part of me found it a very lonely and scary place to be with no one to teach or guide me.

In 1996, experiencing increasing problems, I landed on the doorstep of Dr. Gareth Parry at the University of Minnesota. Not only did Dr. Parry know about HNPP, he was actually studying it! We talked about HNPP. I told him some things that caused pressure palsies in me and he told me more. I still remember sitting in his office and his talking about “rear-end” numbness from sitting too long in the same position. I had struggled for years with movie and airline seats and sitting still at nursing conferences; always warning those I sat next to that I would be squirming a lot. And as Dr. Parry talked about “butt numbness,” I remember this incredulous feeling and thinking “you know that too!” Within minutes, I knew that I was talking to someone who knew a lot more about HNPP than I did in my 11 years of learning on my own. And, miraculously, this man was 5 miles from my house!

I have always been one to grieve my losses and then eventually to try and find meaning and purpose in them. My increasing symptoms meant the end of my nursing career and, at most, working only a few hours a day. I grieved for the loss of my career. For many months I struggled with “Who am I if I can’t do what I’ve trained to do?” And “what do I do with my life now?”

In my talks with Dr. Parry, I learned that HNPP was thought to be as common as CMT1A and there seemed to be no organizations supporting HNPP, nor any information written for the patient. The HNPP void seemed to be a perfect fit with my desire to find purpose in what was happening to me. I knew what it was like to be the only patient doctors had with the dis-



Maureen Horton has been instrumental in increasing the knowledge of HNPP among lay persons and professionals.

ease and to need to self-learn and teach my doctors instead of their teaching me. I had a very strong desire to prevent some of what I had gone through from happening to others, by educating patients. And so, I acted on it.

I cannot speak of what I have done to help the HNPP movement without speaking of the enormous efforts of Dr. Gareth Parry, University of MN, a member of the CMTA Medical Advisory Board and editor of the first CMT handbook (now on sabbatical at Auckland Hospital in New Zealand). Dr. Parry was very interested in HNPP. I think it was his passion. He offered to teach me more about HNPP during his office hours. And I took him up on it, not once, but twice! He offered to edit articles and answer questions that came up. He has been my advisor and mentor. During my personal appointments to see Dr. Parry, for my HNPP, we always ended up talking about HNPP in general. And just talking about HNPP (actually with anyone) would get my enthusiasm flowing and spur me on to do more.

On my own, I tried to contact others with HNPP through the Internet. It took 8 months of searching to find my first person. We developed our own, clumsy, but workable, version of an

Internet support group. Dan Page of Winnipeg, Canada has since taken over as support group monitor and given us a better format. We are now hovering around a worldwide membership of 40 as people come and go. I get excited every time someone new joins and even more excited to hear from other countries. Dr. Parry is our medical advisor and the doctor who answers questions.

My first official efforts, as an advocate, were to contact CMTA (and CMTI) and find out what information they had about HNPP and then to try and get them to take up the HNPP cause. The answer to what information they had was easy. None. So, utilizing information from my talks with Dr. Parry, as well as his editing skills, I created nine pages of patient information about HNPP for the CMTA to send out when people called. CMTA officially recognized HNPP as part of the CMT disorders in the summer of 1997. When I heard the news, I almost jumped out of my skin in excitement. And I phoned family and friends to tell them the news. Since then, one research project on HNPP has been funded, by the CMTA—another milestone. The e-mail support group gives me much information that can be used in articles. And I have tried to have one article in each newsletter.

But just writing a newsletter article was not enough. I wanted people with HNPP to be able to access a lot of information about our disease and to really feel that they are not alone with it. So, I utilized my free web space and created a HNPP web page. I wanted to have a place where people could find basic information about HNPP, ways of treating it, practical advice and tips for living with HNPP and a place to find the elusive knowledgeable doctor to treat people with HNPP.

Doctors Gareth Parry, Richard Lewis at Wayne State, Thomas Bird at the University of

Washington, David Walk at the University of Minnesota and Jim Lupski at Baylor College of Medicine have all helped answer questions. At our own cost (which my husband and I consider part of our donated dollars each year), I have moved the site to obtain an easy web address (www.hnpp.org). The information on the site is slowly building.

Those are my successes to date.

When I was asked to write this article, it was suggested that I write about my failures and frustrations, as well as my successes. As for “failures,” I tend to feel that my timing was not right, but the seed of the idea to bring HNPP more recognition has been planted. Future goals would include individuals with HNPP serving on the CMTA board as well as more physicians getting involved with HNPP. I always feel that my “frustrations” have more to do with my wanting to see things happen quickly. But, it takes time to build a movement of this nature.

Pat Dreibelbis described me as someone who will “not let people ignore HNPP.” The fact that HNPP is new to the CMTA and is slowly increasing its presence is a frustration. I would like to be in a position where HNPP takes on a life of its own and I don’t have to “not let people ignore HNPP.” I would like to see others become more involved, writing more—including people with HNPP, therapists, doctors, the CMTA webmaster and the CMTA staff. I would love to see the language of CMTA publications be more inclusive, and when doctors are interviewed, I would like to see them interviewed for all CMT-related disorders. For HNPP to have more of a presence in both the newsletter and the web page are other steps.

These are my next goals. With time, some effort, and help from others, we WILL get there.

***The West Wing* Introduces MS to the Presidency**

Warner Bros. television series, *The West Wing*, an hour-long drama that gives a behind-the-scenes look at the inner workings of the Oval Office, introduced a new twist to its character of the President, played by Martin Sheen. In an episode this past year, President Josiah Bartlet is found unconscious as he prepares for his State of the Union speech. His condition is initially blamed on the flu, but later in the program, the First Lady discloses that the President actually has multiple sclerosis.

Assigning a disability to any main character of a TV show is still a rare event, particularly if the disability is not meant to be the focus of the character. Portraying a powerful world leader with a disability is an especially positive move by this program.





Members of the CMTA's Medical Advisory Board answer questions from readers.

Ask the Doctor

Dear Doctor,

With very high arches, my orthotics are adjusted every few months and callous build-up is removed. Would you recommend surgery to lower my arches or more dynamic bracing? I walk fairly well, but I am getting some pain in my upper thighs and lower hips.

Would a podiatrist do this type of surgery (lowering the arches). I am 58 years old.

The doctor replies:

Surgery is recommended after physical therapy has been used and adequate braces (AFOs) have been tried. Stretching of the heel cord and plantar fascia can be accomplished early with adequate physical therapy. Foot surgery in patients with CMT needs to be done by an experienced orthopaedic surgeon familiar with the progressive nature of the disease. Different types of surgeries including tendon transfers, osteotomies and triple arthrodesis can be done, but have to be tailored to the patient's deficits and needs. Hip dysplasia is frequent in patients with CMT and needs to be investigated by hip x-rays in all CMT patients who complain of hip pain.



Dear Doctor:

My eldest son inherited CMT from me, but has not been severely handicapped, though he is now 49 years old. He is bothered by the inability to button his shirts and is now taking L-histidine, 500 mg per day and feels it has helped his hand function.

We've been told that this essential amino acid is important for the maintenance of the myelin sheath that protects the nerves. Has any study been done on the positive or negative effect of this acid on CMT patients? Because he is also diabetic, he does not consume much rice or bread, which, I understand, are natural sources of histidine.

I would appreciate any information you can give me. If the histidine is helpful, I will encourage him to continue it. I am aware that it can cause depression in some people and we will watch for that.

The doctor replies:

L-histidine is an essential amino acid that is supplied in adequate amounts in any good

diet. To my knowledge, there are no studies using L-histidine in patients with neuropathies and none in hereditary neuropathies. However, I do not know if L-histidine is beneficial in diabetic patients. If that is the case, he may continue taking it. Diabetes mellitus produces different types of peripheral neuropathies. Some of these are very serious. Your son has two diseases that independently affect the peripheral nerves and in combination can aggravate the nerve damage. The genetic defects in most demyelinating types of CMT are known and the encoded proteins and their functions are already known. However, at the present time there is no effective treatment to stop or slow down the progression of the disease and we do not yet know how to regulate or replace the myelin proteins. There are only symptomatic treatments for CMT, including physical and occupational therapy, bracing and different types of surgeries.

Your son needs the advice of an internist to keep his diabetes under control. He also needs the advice of a neurologist regarding the CMT. A good occupational therapist may recommend some tools to facilitate hand and finger use.



Dear Doctor,

I have been taking Intron-A injected three times a week for a year now for hepatitis C. I am also on Ribavirin, which is an anti-viral, 1000 mg a day for a year. Do you think either or both of these drugs might be responsible for increased symptoms of CMT? They are not on the neurotoxic drug list, but I wonder if other patients are having increased problems while on these medications.

The doctor replies:

Hepatitis C can produce a peripheral neuropathy. I see no adverse reactions to the use of Intron-A, an alfa interferon that seems to interfere with the replication of the hepatitis virus. Ribavirin is any antiviral agent that does not produce an affects in the nerves. Hepatitis C may be a fatal disease; CMT is not. Both medications can give muscle and joint pains.



Dear Doctor,

I have one question. Does CMT affect the spine? I have CMT1X and the spine specialist seems to think that it can.

The doctor replies:

Back pain is a frequent complaint in patients with CMT and is most frequently seen in patients with slapping (steppage) gait. The weak leg muscles put pressure on the back muscles. The symptoms usually improve with the use of AFOs (braces) and anti-inflammatory medications. Kyphosis, or arched back, may be present in some patients.

**Dear Doctor,**

Virtually all of us have been told that there is no treatment for HNPP, although some symptoms can be treated, such as pain. Has anything been tried to treat the HNPP itself, especially for those experiencing more severe symptoms? Or, is everyone assuming that it can't be treated because it is inherited? Could you comment specifically on whether high-dose intravenous immune globulin (IVIg), Avonex, interferon beta-1a, plasmapheresis, or steroids have been tried?

One individual is reporting success with taking Somatropin, a human growth hormone. He claims that with 3 months of treatment, the HNPP symptoms resolved considerably—no numbness, no pins and needles and pain reduced to a mild sensation. When it was stopped, the HNPP symptoms returned over the next 3 to 5 months. Is this a possible treatment for significant HNPP symptoms? Is anyone studying this?

The doctor replies: (Dr. Gareth Parry)

It is true that there is currently no treatment for the disease of HNPP itself. However, there are some potential therapies in the pipeline. Neurotrophic factors are molecules produced in tiny amounts by our own bodies and they have been shown in animals to be able to sustain and stimulate nerves to grow in a variety of neuropathic conditions. So far, none has been successful in humans, but a few trials have been carried out and more are likely in the future. There have been no trials in inherited neuropathies such as HNPP or CMT, but I think it is just a matter of overcoming

some of the technical obstacles. If we can show that these molecules are effective in some other conditions such as diabetic neuropathy, then I think that we may see attempts made to use them in inherited neuropathies.

The other treatments noted (IVIg, Avonex, etc.) are all forms of immunotherapy and have no role in HNPP. A number of patients have received these drugs when it was mistakenly thought that they had a disorder called chronic inflammatory demyelinating polyneuropathy (CIDP), which superficially resembles HNPP. None of them improved as a result of the treatment.

It is interesting that the growth hormone seems to have benefitted one patient. Growth hormone does have neurotrophic activity, although that is not its major action. It suggests that some of the improvement may be related to neurotrophism and supports the concept that the more potent neurotrophic factors may have a role in hereditary neuropathies. However, I would caution everyone against making too much of a single report of subjective improvement.

**Dear Doctor,**

I have never seen the long-term negative effects of walking with foot drop and the typical CMT gait discussed in the newsletter. I know that many doctors recommend bracing to avoid the problems of foot drop and the high steppage gait, but I am unwilling to lose more calf muscle by using braces. What kinds of problems (joint problems?) might I suffer because of my peculiar gait for so many years?

The doctor replies:

CMT is a slowly progressive disease regardless of whether the patient wears braces, has surgery or gets physical or occupational therapy. All of these tools and techniques are necessary and important to treat and control some of the symptoms. At the present time, there is no effective treatment to stop or slow the progression of the disease. The use of adequate bracing improves the patient's gait and prevents muscle cramps due to overuse of weak muscles. Excessive stress and repeated trauma may accelerate the normal wear and tear of joints, the so-called osteoarthritis.

CMTA Support Group News

■ Colorado - Denver Area

Support Group leader, Marilyn Munn Strand, reports that one of her members, Victoria Swain, received an award known as the "9 Who Care" award from a local television station in Denver. They are honoring her for her work with a group called "Newborns in Need." She started the local chapter in Colorado back in 1998 and has been sewing baby clothing and blankets for hundreds of needy babies each month. Her group gives layettes to hospitals and agencies where needy mothers can request them. They also make clothing and blankets for premature babies since they are so small that regular clothing will not fit them. Some of their saddest, but most appreciated, donations are the gowns and outfits they make for stillborn babies so the hospitals will have something nice to dress the baby in for the family to see. They make tiny gowns that fit babies even as small as 1/2 pound. In addition to her sewing, Vickie also writes a monthly newsletter to keep her 100+ volunteers around Denver, and in Kansas, Montana and Wyoming together for the cause. The show honoring Vickie aired on July 22 at 7 PM on Channel 9 in Denver.

■ Missouri/Eastern Kansas

Support group leader and CMTA Vice-President, Ardith Fetterolf, represented the organization at the Neuropathy Association Conference in Branson, Missouri, June 13 through the 18th. Among the presentations was one by Dr. Gil Wolfe, who talked about pain and the drugs being used to assist those with peripheral neuropathies. He mentioned Neurontin as one of the most often prescribed drugs and urged attendees to look at the CMTA's medical alert list regarding drugs that can worsen peripheral neuropathies.

Ardith was one of the presenters and gave a talk on the CMTA, its history, the ongoing research program and the services that the organization offers. At least 12 of the attendees had been diagnosed with CMT, while others were simply told they had idiopathic (of unknown origin) peripheral neuropathy or diabetic neuropathy. Also in attendance was Regina Porter, the support group leader from Oregon.

■ New York - New York City

The new group met for the first time in June and held another meeting for July 15, 2000. Dr.



Ardith Fetterolf, Missouri support group leader, addressed the national meeting of The Neuropathy Association.

David Younger hosted the meeting. The current contact person is Alex Davakis, who can be reached at 212-723-2482. The group meets at the Rusk Institute and would like to encourage new members to call for more information or to join the support group.

■ New York - Westchester County/ Connecticut (Fairfield)

On May 5, 2000, the Westchester County group under leader Kay Flynn, were privileged to have as guest speakers Drs. Victor and Gabi Ionasecu, who have retired to nearby Connecticut. The Ionasecus have long been part of the CMT research community and Victor worked at the University of Iowa prior to his retirement. Both of the Ionasecus were born in Rumania and worked in Bucharest for years. When Victor was invited to join the staff at the University of Iowa, he was forced to leave his wife and two daughters behind. Finally, in 1971 the family was reunited in the United States. It was 1987 before Victor became active in the study of Charcot-Marie-Tooth disorders, when he applied to the MDA for a grant to work on CMT. Since 1987, he has published 25 papers on CMT as well as 152 journal articles. His research has included the discovery of the chromosome 17 flaw in Type IA, the IB defect on chromosome 1 and work with the X chromosome and Connexin32.

The Ionasecus fielded questions from the group members and offered some very personal insight into the CMT research that is ongoing.

CMTA Support Groups

Alabama/Greater Tennessee Valley

Place: ECM Hospital, Florence, AL
Meeting: Quarterly
Contact: William Porter, 205-767-4181

Arkansas—Northwest Area

Place: Harvey and Bernice Jones Center for Families, Springdale
Meeting: 3rd Saturday of each month
Contact: Libby Bond, 501-795-2318
E-mail: charmicoma@netzero.net

California—Berkeley Area

Place: West Berkeley Library
Meeting: Quarterly
Contact: Ruth Levitan, 510-524-3506
E-mail: rulev@pacbell.net

California—Los Angeles Area

Place: Various locations.
Meeting: Quarterly
Contact: Serena Shaffer, 818-841-7763
E-mail: SerenaM71@aol.com

California—Northern Coast Counties (Marin, Mendocino, Solano, Sonoma)

Place: 300 Sovereign Lane, Santa Rosa
Meeting: Quarterly, Saturday, 1 PM
Contact: Freda Brown, 707-573-0181
E-mail: pcmoble@home.com

Colorado—Denver Area

Place: Glory of God Lutheran Church Wheat Ridge
Meeting: Quarterly
Contact: Marilyn Munn Strand, 303-403-8318
E-mail: mmstrand@aol.com

Florida—Boca Raton to Melbourne

Place: Upledger Institute, Palm Beach Gardens
Meeting: Quarterly
Contact: Cynthia Gracey, 561-243-0000

Florida—Miami/Ft. Lauderdale

Place: North Broward Medical Center, Pompano Beach, FL
Contact: Al Kent, 954-742-5200 (daytime) or 954-472-3313 (evenings)
E-mail: marbearwld@aol.com

Kentucky/Southern Indiana/Southern Ohio

Place: Lexington Public Library, Northside Branch
Meeting: Quarterly
Contact: Robert Budde, 859-255-7471

Massachusetts—Boston Area

Place: Lahey-Hitchcock Clinic, Burlington, MA
Meeting: Every other month, the first Tuesday
Contact: David Prince, 978-667-9008
E-mail: baseball@ma.ultranet.com

Michigan—Detroit Area

Place: Beaumont Hospital
Meeting: Three times each year
Contact: Suzanne Tarpinian, 313-883-1123

Michigan—Flint

Place: University of Michigan, Health Services
Meeting: Quarterly
Contact: Debbie Newberger/Brenda Kehoe, 810-762-3456

Minnesota—Benson

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

Mississippi/Louisiana

Place: Clinton Library, Clinton, MS
Meeting: Quarterly
Contact: Betty Aultman, 601-825-5626
 Julia Provost, 601-825-6482

Missouri/Eastern Kansas

Place: Mid-America Rehab Hospital, Overland Park, KS
Meeting: First Saturday bi-monthly
Contact: Lee Ann Borberg, 816-229-2614
E-mail: ardi5@aol.com

Missouri—St. Louis Area

Place: St. Louis University Medical Health Ctr.
Meeting: Quarterly
Contact: Carole Haislip, 314-644-1664

New York—New York City

Place: Rusk Institute
Meeting: Bimonthly
Contact: Alex Davakis, 212-723-2482

New York—Horseheads

Place: NYSEG Meeting Room, Rt. 17
Meeting: Quarterly
Contact: Angela Piersimoni, 607-562-8823

New York (Westchester County)/Connecticut (Fairfield)

Place: Blythedale Hospital
Meeting: Monthly, Saturday
Contact: Kay Flynn, 914-793-4710
E-mail: alma622@worldnet.att.net

North Carolina—Archdale/Triad

Place: Archdale Public Library
Meeting: Quarterly
Contact: Ellen (Nora) Burrows, 336-434-2383

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

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

Ohio—Greenville

Place: Church of the Brethren
Meeting: Fourth Thursday, April-October
Contact: Dot Cain, 937-548-3963

Oregon—Willamette Valley

Place: Brooks Assembly of God Church
Meeting: Monthly
Contact: Regina Porter, 503-591-9412
 Maryann DiStefano-Hill, 503-585-3341
E-mail: moonglow21@aol.com

Texas—Dallas/Ft. Worth

Place: Harris Methodist HEB Hospital
Contact: Greta Lindsey, 817-281-5190 or Shari Clark, 817-543-2068
E-mail: jdsbclark@webtv.net

OF INTEREST

The Archives of Neurology have several offerings of interest to CMT patients. In Vol. 57, No. 3, Medical Advisory Board (MAB) Member, Thomas D. Bird, M.D. has written an editorial about testing in genetic diseases called, "Sporadic Cases of Possible Genetic Diseases: To Test or Not to Test?" In the May issue, another MAB member, Jeffrey M. Vance, Ph.D., M.D. published an editorial entitled, "The Many Faces of Charcot-Marie-Tooth Disease." In the same issue, an article discusses how "Hereditary Motor Sensory Neuropathy Type 2C is Genetically Distinct from Types 2B and 2D." Going back to old issues, in July 1999, an observation was published that CMT 2C is characterized by axonal neuropathy, diaphragm weakness and vocal cord paralysis in a group of kindred.



BIBLIOGRAPHY OF MAB PUBLICATIONS, 1999-2000 (Continued from page 7)

- Mouton P, Tardieu S, Gouider R, Birouk N, Maisonnobe T, Dubourg O, Brice A, LeGuern E, Bouche P.** "Spectrum of clinical and electrophysiologic features in HNPP patients with the 17p11.2 deletion." *Neurology*. 1999 Apr 22; 52(7):1440-6.
- Nelis E, Haites N, Van Broeckhoven C.** "Protein Mutations in the peripheral myelin genes and associated genes in inherited peripheral neuropathies." *Human Mutation*. 1999; 13(1):11-28.
- Nelis E, Timmerman V, De Jonghe P, Van Broeckhoven C, Rautenstrauss B.** "Molecular genetics and biology of inherited peripheral neuropathies: a fast-moving field." *Neurogenetics*. 1999 Sep; 2(3):137-48.
- Neuberg DH, Suter U.** "Connexin32 in hereditary neuropathies." *Advances in Experimental Medical Biology*. 1999; 468:227-36.
- Nicholson GA.** "Mutation testing in Charcot-Marie-Tooth neuropathy." *Annals of the New York Academy of Science*. 1999 Sep 14; 883:383-8.
- Niemann S, Sereda MW, Rossner M, Stewart H, Suter U, Meinck HM, Griffiths IR, Nave KA.** "The 'CMT rat': peripheral neuropathy and dysmyelination caused by transgenic overexpression of PMP22." *Annals of the New York Academy of Science*. 1999 Sep 14; 883:254-61.
- Othmane KB, Johnson E, Menold M, Graham FL, Hamida MB, Hasegawa O, Rogala AD, Ohnishi A, Pericak-Vance M, Hentati F, Vance JM.** "Identification of a new locus for autosomal recessive Charcot-Marie-Tooth disease with focally folded myelin on chromosome 11p15." *Genomics*. 1999 Dec 15; 62(3):344-9.
- Pareyson D, Menichella D, Botti S, Sghirlanzoni A, Fallica E, Mora M, Ciano C, Shy ME, Taroni F.** "Heterozygous null mutation in the P0 gene associated with mild Charcot-Marie-Tooth disease." *Annals of the New York Academy of Science*. 1999 Sep 14; 883:477-80.
- Potocki L, Chen KS, Koeuth T, Killian J, Iannaccone ST, Shapira SK, Kashork CD, Spikes AS, Shaffer LG, Lupski JR.** "DNA rearrangements on both homologues of chromosome 17 in a mildly delayed individual with a family history of autosomal dominant carpal tunnel syndrome." *American Journal of Human Genetics*. 1999 Feb; 64(2):471-8.
- Potocki L, Chen KS, Park SS, Osterholm DE, Withers MA, Kimonis V, Summers AM, Meschino WS, Anyane-Yeboah K, Kashork CD, Shaffer LG, Lupski JR.** "Molecular mechanism for duplication 17p11.2- the homologous recombination reciprocal of the Smith-Magenis microdeletion." *Nature Genetics*. 2000 Jan; 24(1):84-7.
- Reiter LT, Liehr T, Rautenstrauss B, Robertson HM, Lupski JR.** "Localization of mariner DNA transposons in the human genome by PRINS." *Genome Research*. 1999 Sep; 9(9):839-43.
- Sahenk Z, Mendell JR.** "Alterations in nodes of Ranvier and Schmidt-Lanterman incisures in Charcot-Marie-Tooth neuropathies." *Annals of the New York Academy of Science*. 1999 Sep 14; 883:508-12.
- Sancho S, Magyar JP, Aguzzi A, Suter U.** "Distal axonopathy in peripheral nerves of PMP22-mutant mice." *Brain*. 1999 Aug; 122 (Pt 8):1563-77.
- Scherer S.** "Axonal pathology in demyelinating diseases." *Annals of Neurology*. 1999 Jan; 45(1):6-7.
- Scherer SS, Bone LJ, Deschenes SM, Abel A, Balice-Gordon RJ, Fischbeck KH.** "The role of the gap junction protein connexin32 in the pathogenesis of X-linked Charcot-Marie-Tooth disease." *Novartis Foundation Symposium*. 1999; 219:175-85; discussion 185-7.
- Scherer SS, Fischbeck KH.** "Is CMTX an axonopathy?" *Neurology*. 1999 Jan 15; 52(2):432-3.
- Shy ME, Kamholz J, Lovelace RE.** "Introduction to the Third International Symposium on Charcot-Marie-Tooth disorders." *Annals of the New York Academy of Science*. 1999 Sep 14; 883:xiii-xviii.
- Stogbauer F, Young P, Kuhlenbaumer G, Kiefer R, Timmerman V, Ringelstein EB, Wang JF, Schroder JM, Van Broeckhoven C, Weis J.** "Autosomal dominant burning feet syndrome." *Journal of Neurosurgery and Psychiatry*. 1999 Jul; 67(1):78-81.
- Stronach EA, Clark C, Bell C, Lofgren A, McKay NG, Timmerman V, Van Broeckhoven C, Haites NE.** "Novel PCR-based diagnostic tools for Charcot-Marie-Tooth type 1A and hereditary neuropathy with liability to pressure palsies." *Journal of the Peripheral Nervous System*. 1999; 4(2):117-22.
- Suter U, Nave KA.** "Transgenic mouse models of CMT1A and HNPP." *Annals of the New York Academy of Science*. 1999 Sep 14; 883:247-53.
- Thomas FP, Geller TJ, Hahn AF, Gutmann L, Huang XL, Wu H, Wyandt HE, Lebo RV.** "Modification of CMT1 phenotypes by the independent coexisting neurogenetic disorders, McArdle disease and chromosome 5p trisomy." *Annals of the New York Academy of Science*. 1999 Sep 14; 883:472-6.
- Thomas PK.** "Overview of Charcot-Marie-Tooth disease type 1A." *Annals of the New York Academy of Science*. 1999 Sep 14; 883:1-5.
- Timmerman V, Beuten J, Irobi J, De Jonghe P, Martin JJ, Van Broeckhoven C.** "Distal hereditary motor neuropathy type II (distal HMN type II): phenotype and molecular genetics." *Annals of the New York Academy of Science*. 1999 Sep 14; 883:60-4.
- Timmerman V, De Jonghe P, Ceuterick C, De Vriendt E, Lofgren A, Nelis E, Warner LE, Lupski JR, Martin JJ, Van Broeckhoven C.** "Novel missense mutation in the early growth response 2 gene associated with Dejerine-Sottas syndrome phenotype." *Neurology*. 1999 Jun 10; 52(9):1827-32.
- Timmerman V, De Jonghe P, Van Broeckhoven C.** "Report of the 8th annual symposium of the European Charcot-Marie-Tooth Consortium, Antwerpen, Belgium, July 2-4, 1999." *Journal of the Peripheral Nervous System*. 1999; 4(3-4):279-304.
- Vance JM.** "Charcot-Marie-Tooth disease type 2." *Annals of the New York Academy of Science*. 1999 Sep 14; 883:42-6.
- Warner LE, Garcia CA, Lupski JR.** "Hereditary peripheral neuropathies: clinical forms, genetics, and molecular mechanisms." *Annual Review of Medicine*. 1999; 50:263-75.
- Warner LE, Svaren J, Milbrandt J, Lupski JR.** "Functional consequences of mutations in the early growth response 2 gene (EGR2) correlate with severity of human myelinopathies." *Human Molecular Genetics*. 1999 Jul; 8(7):1245-51.

Letters to the Editor:

Dear CMTA,

I noticed that the ToeOff (mentioned in the Autumn 1999, CMTA Report) is a 9-oz. brace different from the ones I have seen previously. There is a foot piece that fits between the shoe and the insole and stays in the shoe, unless you are changing shoes. An orthotist recommended this for me. She says it will only work if there is side-to-side stability, which I still have. By bracing the front of the leg, one can undo the knee strap and be able to drive easier (pushing down on the pedal and not going against the AFO).

The brace is metal. When you walk, you are bending it some. The metal tries to go back to its original position and in doing so, gives a bit of spring action to the step.

—M.H., MN

Dear CMTA,

My brother-in-law has CMT disease and would like to correspond with others who have this disease. Is there any way he could get others to write to him about the condition or just for support? His name and address is: Mr. Bernard Shova, HCR 1, Box 1728, Malone, NY 12953.

He would appreciate hearing from others with the disease.

—P.S., NY

Dear CMTA,

First of all, I want to thank you because you gave me the opportunity to correspond with your association, which has been so helpful to me. I was very troubled before I found you. I didn't know anything about this peripheral neuropathy because the doctors never explained it to me. I didn't know about the toxic drugs and many other things.

As I told you in my last letter, four members of my family have this disease. I think you are a message that the angels sent to me. Many questions trouble me...what occupation will my children be able to have? They have graduated from high school and plan to pursue their studies at a university in Greece to become useful and knowledgeable people. When I was young, I was a prima ballerina. My dream was that my two girls would become ballerinas like me, but my dream has gone by. I never imagined that they would have the misfortune to be born with this progressive, neuromuscular disease.

Now that I'm 36 years old, I finally understand that we will become what God wants us to become and not necessarily what we want. Still, where there is a will, there is a way and we must keep trying our whole lives.

My husband and my children are Type I, as our doctor said. We have not had the DNA blood test yet because it is so expensive and we can't cover the cost. All the members of my family who have CMT have high arches and claw toes. I'm not sure if my husband should be wearing AFOs. Many times I've thought I must move from my country and come closer to live near you because of this problem that I face with my whole family. But, it is not really a serious thought. I'm sure you will help me all you can even though we are far apart.

I'm enclosing my membership and the money for the special offer of the Facts series. Across this long way, I'm sure we'll find and discover useful help and things we can do to help each other.

—K.S., Greece

Dear CMTA,

Having inherited CMT, I am interested in reading about the fantastic breakthroughs in genetic research rapidly occurring. On Wednesday, June 27, our Phoenix paper carried the news of the mapping of the human genome.

I thought you should see how Phoenix's St. Joseph's Hospital is involved, through researcher Kumaraswami Sivakumar. The paper referred to his work: "Researcher Kumaraswami Sivakumar, for example, is using genetics to look at Charcot-Marie-Tooth, a common inherited nerve disease that weakens and atrophies leg muscles. Researchers are hopeful that if they can locate the one protein, a possible byproduct of a malfunctioning gene, that causes the disorder, they could treat it."

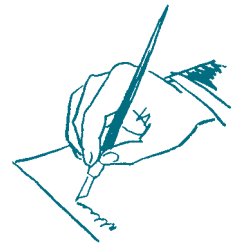
I have not officially been tested for a specific type of CMT, but am at mid-age and am making large adjustments in my lifestyle because of CMT.

—S. H., AZ

Dear CMTA,

Your Spring 2000 Report was great! Lots of useful information and I especially liked the peripheral nerve drawings. The drawings make the subject matter very easy to understand.

—B.B., OK



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.

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

MEDICAL ALERT:

These drugs are toxic to the peripheral nervous system and can be harmful to the CMT patient.

Adriamycin
Alcohol
Amiodarone
Chloramphenicol
Cisplatin
Dapsone
Diphenylhydantoin (Dilantin)
Disulfiram (Antabuse)
Glutethimide (Doriden)
Gold
Hydralazine (Apresoline)
Isoniazid (INH)
Megadose of vitamin A*
Megadose of vitamin D*
Megadose of vitamin B6* (Pyridoxine)
Metronidazole (Flagyl)
Nitrofurantoin (Furadantin, Macrochantin)
Nitrous oxide (chronic repeated inhalation)
Penicillin (large IV doses only)
Perhexiline (Pexid)
Taxol
Vincristine
Lithium, Misomidazole, and Zoloft can be used *with caution.*

Before taking any medication, please discuss it fully with your doctor for possible side effects.

*A megadose is defined as ten or more times the recommended daily allowance.



What is CMT?

- ... is the most common inherited neuropathy, affecting approximately 150,000 Americans.
- ... may become worse if certain neurotoxic drugs are taken.
- ... can vary greatly in severity, even within the same family.
- ... can, in rare instances, cause severe disability.
- ... is also known as peroneal muscular atrophy and hereditary motor sensory neuropathy.
- ... is slowly progressive, causing deterioration of peripheral nerves that control sensory information and muscle function of the foot/lower leg and hand/forearm.
- ... causes degeneration of peroneal muscles (located on the front of the leg below the knee).
- ... causes foot-drop walking gait, foot bone abnormalities, high arches and hammer toes, problems with balance, problems with hand function, occasional lower leg and forearm muscle cramping, loss of some normal reflexes, and scoliosis (curvature of the spine).
- ... does not affect life expectancy.
- ... has no effective treatment, although physical therapy, occupational therapy and moderate physical activity are beneficial.
- ... is sometimes surgically treated.
- ... is usually inherited in an autosomal dominant pattern, which means if one parent has CMT, there is a 50% chance of passing it on to each child.
- ... Types 1A, 1B, 1X, and HNPP can now be diagnosed by a blood test.
- ... is the focus of significant genetic research, bringing us closer to answering the CMT enigma.

The CMTA Report

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



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

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