Charcot-Marie-Tooth Association

THE CMTA REPORT

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CMTA Medical Advisory Board Announces Recent Research Findings

By Roger Lebo, Ph.D.

The MAB of the CMTA met in April at the meetings of the American Academy of Neurology. Dr. Robert E. Lovelace of Columbia Medical Center is the Chairman of the MAB. Dr. Roger Lebo of the University of California at San Francisco was the organizer and moderator of the meeting. Over twenty clinical and research scientists attended the meeting and discussed current CMT research. The following is Dr. Lebo's report of the seminar.

Medical Advisory Board meeting attendees

The Charcot-Marie-Tooth Association sponsored its third annual satellite meeting at the American Academy of Neurology Conference in New York City on April 27, 1993. Dr. Phillip Chance reported a neuropathy he discovered that is the alternative mutation to CMT IA, and Dr. Jeffrey Vance reported further evidence of additional autosomal recessive CMT categories. These results further define the multiple genetic origins of CMT. (Fig. 1) An outline of positional cloning was presented to clarify the research status of each CMT disease with the most significant milestones indicated (Fig. 2). Positional cloning was completed for CMT IA when mutations in the peripheral myelin protein (PMP22) gene were reported independently by Dr. Valentijn and Dr. James Lupski. Other positional cloning investigations are proceeding toward isolating additional CMT genes. Dr. Chance summarized the status of CMT IA.

CMT IA often results from unequal recombination between normal chromosomes generating one daughter chromosome with duplicate genes and the other with deleted genes; both types of chromosomes result in peripheral neuropathy. The most common form of CMT (CMT IA) mapped to chromosome 17 by Dr. Vance was found to result from a duplication of about 1% of that chromosome. This is an autosomal dominant genetic disease that is passed from one affected parent to his/her children. Each time an affected parent has a child, that child has a 50 - 50 chance of inheriting CMT. At the last CMTA satellite meeting a candidate gene (Peripheral Myelin Protein 22;PMP22) was mapped to this duplicated region after an abnormal PMP22 gene was shown to cause a similar disease in mice. Since then, point mutations in the human PMP22 gene have been found. This confirms that is at least one of the genes, if not the only gene

(continued on p.2)

IN THIS ISSUE:

Recent Research Findings - cont'd

in the duplication that results in CMT1A. About half of all CMT patients and two thirds of CMT patients with slow nerve conduction have CMT1A. Nearly all patients with CMT1A have a duplication in the CMT1A region. A majority of these mutations are the same size but up to one third have smaller duplications with similar CMT symptoms. The largest duplication region has been isolated in DNA clones and mapped.

Dr.Chance described Hereditary Neuropathy with Liability to Pressure Palsy (HNPP) which he published in CELL. He found about 1% of chromosome 17 is deleted from the CMT 1A gene region in two families with (HNPP). This the reverse mutation of the commonly occurring CMT1A duplication. Like CMT 1A, HNPP affects peripheral nerves emanating from the spinal card. However, a significant difference in HNPP includes slow recovery of nerve function after pressure is applied.

Dr. Victor Ionasescu presented the current status of the consortium working on the X chromosome form of CMT (CMTX). The gene on band Xq13 has cosegregated with phosphoglycerate kinase 1 gene in every informative case so that these two genes are in the same subchromosomal region. Dr. Ionasescu also discussed an unusual CMT1A family with some members that have no detectable duplication and other members that have a duplication. One explanation is that a small undetected duplication in one ancestor with the disease resulted in a second chromosome error producing a larger detectable duplication.

Dr.Vance and his postdoctoral fellow Dr. Kamel Ben Othmane from Tunisia, identified three different forms of autosomal recessive CMT based upon different nerve pathophysiology observed under the microscope. Autosomal recessive CMT results when a child inherits one abnormal copy of the same gene from each parent. Because marrying a first cousin is common in some cultures (called inbreeding), two copies of the some abnormal gene are passed to children from these marriages more often. Thus recessive CMT is seen more often in large Tunisian familia which provide an ideal patient population to study these genes. Linkage analysis with these patients is further defining these gene locations. Dr. Vance presented other progress in linkage studies to locate the autosomal dominant CMT2 locus in large families. CMT2 affects the nerve cell (axon) that conducts a nerve impulse from the brain to the muscle, while CMT1 disassembles the myelin in the Schwann cell that insulates the nerve impulse from surrounding tissue as it travels to the muscle.

Dr. Robert Lovelace, Columbia University, presented his findings that pregnancy in CMT patients can result in significantly accelerated disease progression. Based upon his initial sampling of 20 patients, 12 patients had significant functional degeneration in the second or subsequent pregnancy. These preliminary findings suggest that pregnant CMT patients should be advised of this risk and monitored regularly so that physical therapy can be initiated as indicated.

Dr. Lebo reported the results of a prenatal test that diagnoses CMT1A duplication by multicolor in situ hybridization. Previously his laboratory developed this method for another genetic disease that results from a gene deletion. Although gene duplication can be detected by other methods, in situ hybridization is either faster, informative more often, and/or

(concluded on p. 3)

Call for Articles

The CMTA Report welcomes your ideas and article suggestions. For example, you may submit a human interest story telling of your experience of living with CMT. Also, medical professionals can forward articles of a clinical or medical nature that would be of general interest to our readership.

CMTA Medical Advisory Board

CMT1A Test Diagnoses At Risk Fetuses & Patients

Dr Roger Lebo, University of California-San Francisco, developed a test to diagnosis gene and aneuploidy using multi-color flourescence in situ hybridization. After applying this protocol to another human genetic disease, steroid sulfatase deficiency, Dr. Lebo used it to test for the CMT1A gene duplication.

His study found at least three different size duplications in the CMT1A gene region. In situ hybridization detected one-third more duplications than restriction enzyme analysis with the previously recommended best probe.

If the CMT1A duplication on chromosone 17 is found in an affected relative, then both at risk fetuses and other family members can be tested for CMT. Thus far this test has diagnosed 6 of 6 fetuses using samples obtained routinely by both amniocentesis and chorionic villus sampling. A small whole blood sample is needed for diagnosing CMT in all other suspected patients.

This test costs $240 for each subject tested. For more information contact Dr Lebo at Room U-253, University of California 533 Parnassus Avenue San Francisco CA 94143-0720 or call (415) 476-5481.

Editors Note: References for Dr. Lebo's work are available from the CMTA upon request.

Recent Research Findings - cont'd

can be done with smaller samples than these other methods. Prenatal diagnosis of CMT1A is best done by fluorescence in situ hybridization. The CMT parent of an at-risk fetus is first tested for the CMT1A duplication. Then fetuses of mothers or fathers with the duplication are diagnosed using samples obtained by amniocentesis or chorionic villi sampling. This approach also diagnoses patients who are at-risk for CMT1A but who have not shown any symptoms.

Dr. Lovelace concluded the meeting by presenting plans for the Third International Charcot-Marie-Tooth Disease Conference in 1995.

Editor's note: For copies of Dr. Lebo's charts (Fig. 1 & Fig. 2) send a SSAE to the CMTA requesting them.

CMT Gene Identified

Dr. James Lupski

Dr. James R. Lupski, Baylor College of Medicine, and his group in collaboration with the groups of Dr. Pragna Patel, of Baylor College of Medicine, and Dr. Eric Shooter, of Stanford University, have identified a gene responsible for Charcot-Marie-Tooth disease. Their work was published in the July 8, 1993 issue of The New England Journal of Medicine. In this paper they demonstrate that a patient with a spontaneous mutation in the peripheral nerve specific gene PMP22 has the CMT disease phenotype. (Editor's note: Phenotype means the patient physically exhibits the symptoms of CMT.)

The path to the identification of the CMT gene has been long and arduous. Initially, several years were spent collecting data from large families with autosomal dominantly inherited CMT and using positional cloning efforts to identify the CMT locus. Dr. Lupski's and Dr. Patel's groups jointly published the discovery in 1991 that CMT 1A appeared to be associated with a large DNA duplication. It was not known if there was only one gene or several genes contained within this large inherited DNA rearrangement that was responsible for the disease phenotype. In 1992 the group published that patients with partial trisomy of the region had the same electrophysiological findings as patients with inherited CMT, suggesting that gene dosage was important to the CMT phenotype. In mid 1992 several groups from around the world demonstrated that the peripheral nerve specific gene PMP22 mapped within the CMT 1A DNA duplication. The critical finding then was to identify patients that did not have the CMT 1A duplication and screen their PMP22 gene. These results were reported in The New England Journal of Medicine, wherein Dr. Benjamin Roa, working in Dr. Lupski's laboratory, identified a spontaneous PMP22 mutation associated with the onset of CMT in a family. A different point mutation in the PMP22 gene has also been identified in inherited CMT by a Dutch group.

This work on CMT has important implications for human genetic diseases. It clearly demonstrates that for one of the most common dominant disorders in man, there is no mutant gene, but instead the disease appears to occur from having three copies of a normal gene. Thus, gene dosage effects can be very important for dominantly inherited traits. Moreover, the work on CMT blurs the boundaries between single gene disorders that segregate in the Mendelian fashion and cytogenetic disorders which have DNA rearrangements such as duplications or deletions.

Genica Pharmaceuticals Announces CMT Blood Test

Genica Pharmaceuticals Corp., a biotechnology company specializing in diagnostics and therapeutics for neuromuscular and neurological diseases, announced July 7, 1993 that the first commercially available blood test to diagnose CMT or detect the presence of the gene that causes the disease in people who might pass it on to their children, will be available in August. The test uses a technology known as pulsed-field gel electrophoresis (PFGE). This test is specific for type IA CMT which is caused by the CMT gene located on chromosome 17. (See cover story.) The cost of the test is $395.00. If you are interested in this diagnostic test your physician may obtain a shipping kit complete with instructions and a pre-paid air bill by calling Sarah Quiry, a Genica customer service representative, at 800-394-4493 ext. 105. If you have technical questions please direct your call to Claudia Soltys at ext. 106. All that is necessary for the test is two samples of whole blood collected in yellow top tubes (ACD solution A). The results will be sent to your physician in three weeks.

ITEMS of INTEREST

ITEM 1: The Charcot-Marie-Tooth Association announces a regional meeting for patients, family and physicians on September 18,1993 at the Sheraton South Colorado Springs Hotel. The meeting organizer is Dr. Greggory Stilwell, CMTA support group leader. The keynote speaker will be Dr. Robert Sampson discussing, "Surgical and other Therapeutic Approaches to CMT Foot/Ankle Deformities." There will also be a panel discussion on "Salvage Procedures for Past Failed Surgery." Bring your x-rays. Other presentations include Dr. David Labosky on "Hand Deformities:Bracing vs. Surgery, Dr. Greg Stilwell on "The Use of Magnetic Resonance Imagining for Calf Muscle Rehab and Tendon Transfer Planning" and Dr. Emory Cowan on "Coping with Psychological Aspects of Chronic Disease." For registration and information, contact:

Marjorie Cowan
5317 Crackerbarrel Circle
Colorado Springs, CO 80917
719-570-7844

ITEM 2: There is a new catalogue available which has high quality items such as an electric jar opener, a lever action bottle opener and many home products with adaptable grips. The catalogue is called Good Idea!, P.O. Box 5111, Englewood, CO 80155 or call toll free 1-800-538-6690.

ITEM 3: NO PITY: People with Disabilities Forging a New Civil Rights Movement is the first book to tell the history and ongoing struggles of this powerful, new civil rights movement. Written by U.S. News and World Report editor Joe Shapiro, NO PITY brings to life the issues, politics and people that have shaped the disability rights movement over the last three decades, from its inception at Berkeley in the 1960's, through the enactment of the Americans with Disabilites Act to issues that will face people with disabilites in the future. There are 43 million disabled Americans, many of whom were not born with their disability. It is the one minority that anyone can join at any time, as the result of a sudden automobile accident, a fall down a flight of stairs, cancer or disease. NO PITY forcefully demonstrates that disability rights is an issue that affects all Americans. The Americans with Disabilities Act of 1992 may have passed in Congress, but the struggle to overcome society's myths, fears, and prejudices is far from over. People with disabilities need equal opportunity and access, not pity. Order NO PITY (Times Books;May 19,1993;$ 25.00) by calling toll free 1-800-733-3000.

ITEM 4: The Advocacy Center of Tallahassee, Florida offers the following: Ten Steps to Being an Effective Self-Advocate

1. Believe in Yourself: You are worth the effort it takes to protect your interests and your rights.You can do it!
2. Realize You Have Rights: You are entitled to equality under the law. Inform yourself by using the resources and asking questions. Insist that explanations are clear and understandable. Remember, service providers are public servants. They work for you!
3. Discuss Your Concerns: Talk directly with your service provider either by phone, in person or write a letter. You may bring someone along for support.
4. Get The Facts: Problem solve by being informed. Get the facts in writing. Ask for the policies or the regulations being cited to you. People sometimes settle for a quick verbal decision that may not be accurate. Hold agencies accountable for the decisions they make.
5. Use the Chain of Command: Use an agency's chain of command to make sure a supervisor or someone else with authority has an opportunity to work with you on the problem and resolution.
6. Know Your Appeal Rights: Request clear information on your appeal rights within the agency, and outside the agency. Know what the next step will be if you are dissatisfied.
7. Be Assertive and Persistent: Keep after what you want. Remember that effort moves bureaucracies. Follow up!
8. Use Communication Skills: Have a plan outlining your concerns. Stay calm and express yourself clearly. Be willing to listen because what you hear may be as important as what you say.
9. Ask For Help: Link up with advocacy organizations for more specific information on problems you are having obtaining services related to a disability. Remember there are also community support groups or organizations.
10. Follow Up: Don't give up without using these skills. Agencies are accountable for the decisions they make. You are entitled to know and exercise all your options to obtain the assistance you need.

ITEM 5: Don't forget that CMT FACTS II is now available. This volume along with CMT FACTS I should be in every patient's personal library. Order forms appear in the back of this issue.

ITEM 6: Available from NARIC and ABLEDATA is a fact sheet called Funding Assistive Technology, Number 14, July 1992. This eleven page fact sheet has explanations of the procedures one might have to go through as well as agencies and organizations that might be able to help a person get funding for such items as ambulation aids, mobility aids, prosthetics and orthotics, safety aids, and communication aids. The new number for ABLEDATA is 800/346-2742.

ITEM 7: A powered mobility device can make a big difference in how efficiently a person can get around in the environment and how much energy or time he or she has available for home, community, and work activities. Powered scooters are a simpler and less expensive option than traditional power wheelchairs for people who don't require sophisticated electronics or the seating and control options now available on some wheelchairs. Many people who use powered scooters have some ability to walk but need the device for longer distances, energy conservation, or at times when their walking ability is more impaired. Scooters usually require good upper body and arm function. For a complete discussion of scooters, send for Fact Sheet "Powered Scooters" # 5 from ABLEDATA, 800/346-2742.

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The Mayo Clinic Dynamine Study: A Personal Account

by Diana Eline

Dynamine, 3,4, diamino pyridine, has previously been shown to be very effective in Lambert Eaton Syndrome, a hereditary form of myasthenia gravis, by Drs.Windebank, McCavoy and Russel at the Mayo Clinic. (Dynamine acts as a potassium channel blocker which potentially would prolong the length of the nerve cell action potential and thereby increases the rate of nerve conduction.) It was hypothesized that the drug might be effective in CMT and other demyelinating neuropathies as it would, hopefully, increase the rate of conduction in the diseased nerves and thereby cause clinical improvement through better muscle function.

Approximately 46 patients with CMT Type I were invited to the Mayo Clinic to participate in the clinical trials of Dynamine. The study required two weeks of stay at the Mayo Clinic. One week the patient was on the drug and one week they were on the placebo, not knowing which week was which. It was a double blind study in which neither the physician who performed the daily neurological exams nor the patient knew whether the drug or the placebo was being administered.

The patients were warned ahead of time about the potential side effects. I believe that I was on the drug during the second week because of the side effect of numbness and tingling around my mouth starting about 45 minutes after I took the dose on the third day of the drug administration. This was the day the dose was increased to 20 mgs. Dynamine is a drug that is theoretically only effective while it is in the bloodstream. It clears the body within 5 to 6 hours. The dose was given every 4 hours during the day in order to maintain blood levels.

I do not believe that I experienced any dramatic changes in motor function while on the drug, nor did the other CMT Type I patients that I met who were in the study and had completed their second week. However, my sensation seemed to have gotten stronger. I wasn't looking for a change in sensation; but, in the morning of the third day, when the dose was raised to 20 mgs, I put my hands under running cold water and the sensation felt stronger than I ever remember. Of course, I would much rather have experienced an improvement in motor function.

Only when the results of the tests, including the EMGs, the biomechanical tests and the daily neurological exams, are analyzed will a true determination be made as to whether or not I benefitted from the drug. If I did, I would be given the option of being on the drug for a year, and would have to return to the Mayo Clinic once a month for the first 3 months.

I must give a disclaimer that this is just my experience and that of two other patients that I met. I was given permission by Dr. Russell to discuss my experience as the study is near completion and he has received all the volunteers needed. Perhaps it will be determined later that Dynamine was effective for other CMT patients or a subset of them. The definitive conclusions can only be drawn after the paper is submitted for publication, which Dr. Russell tells me should be within a year or so. Dr. Russell and the nurses in the clinical research center alluded to the fact that the results on the CMT patients will, in all probability, not be as dramatic as those seen in the Lambert Eaton patients.

(Editor's Note: This is the first drug study done on CMT patients since 1986 when Dr. Bradley published his results on the drug, Cronasssial.)

Referrals Available

The CMTA has compiled a list of neurologists, orthopedists, physiatrists (a physiatrist is a physician trained in physical medicine and rehabilitation) and podiatrists who have a special interest in CMT. We can also access respiratory specialists. Additionally, we have listings for pedorthists. A pedorthist is a practioner who provides care to the patient by fitting orthopedic shoes and devices, at the direction of and in consultation with physicians.

To receive any of these referrals send a stamped self-addressed business-sized envelope indicating the geographic areas needed to: CMTA, 601 Upland Avenue, Upland, PA 19015.

For referrals for a hand surgeon contact the American Society for Hand Surgery, 3025 South Parker Road, Suite 3025, Aurora, CO 80014, 303/755-4588.

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Dietary Supplements

Editor's note: This article is reprinted with permission from the October 1992 issue of the NORD ON-LINE Bulletin. We are continually being asked about food and vitamin therapies for CMT. Nutritionists and physicians have consistently maintained that a well balanced, nutritionally sound diet and weight control are necessary for the CMT patient. There is no known accepted diet therapy or diet supplement for the treatment of CMT. The information in this article points out some of the problems with dietary supplements and vitamins.

NORD is very concerned about dietary supplements. Because these supplements are natural components of foods, they are not patentable. Therefore, pharmaceutical manufacturers will not develop them as "drugs' for medical purposes. Dietary supplements are usually sold in health food stores and are manufactured by many companies that are not regulated by the Federal Drug Administration (FDA). There are valid medical uses for some dietary supplements, but the unreliability of health food products means physicians cannot trust them for potency and bioavailability.

Certain vitamins, minerals and other dietary supplements have valid medical uses for certain inborn errors of metabolism. Healthy people, however, do not need these supplements if they eat a balanced diet. Yet the health food business is a multi-billion dollar industry which skirts the law by providing supplementary information (usually brochures and magazine advertisements) which often make unscientific and misleading medical claims about these products. As long as the medical claims are not printed on the products label, there is nothing FDA can do to stop a manufacturer from selling a product as "nutritional supplement" even though it is marketed surreptitiously as a drug.

People who buy health foods to treat a medical condition often forgo seeing a doctor and they are not aware of possible side effects because this information is omitted from literature which talks about only the benefits the product will provide. Outrageous claims are made about the medical benefits of these products which often target cancer, arthritis, impotence, baldness, and any condition deemed lucrative to the industry.

Because certain metabolic diseases are treatable with nutritional supplements, and because some of these products are available only through health food stores, NORD sent a number of these nutritional supplements to Duke University for testing. We wanted to see whether consumers are getting what they pay for when they purchase these products from a health food store. In one case we compared an FDA approved prescription drug, L-carnitine (Carnitor), with versions of carnitine that are sold as nutritional substances without FDA monitoring. We particularly chose carnitine because some health insurance companies and state Medicaid systems have denied reimbursement for the prescription version, Carnitor, telling patients they can buy the less expensive non-prescription product as a nutritional supplement at health food stores.

> ... seven of the twelve brands of carnitine do not disintegrate in the stomach, therefore they have no medical benefit to patients.

The study found that seven of the twelve brands of carnitine (eleven of which are sold in health food stores) do not disintegrate in the stomach, therefore they have no medical benefit to patients. There was a wide variance of bioavailability between specimens taken from the same bottle, illustrating that there is a lack of quality control in the manufacturing of these health food products. Moreover, many of the capsules and tablets in the non-prescription versions of L-carnitine delivered 60% or less of the active ingredient as listed on their labels; one brand contained no L-carnitine at all; another brand contained a barely detectable amount of L-carnitine.

Carnitine levels were lower than the dose listed on the label of most health food products either because they failed to disintegrate or because they contained less carnitine than they stated. The prescription version of the drug, which is regulated by the FDA, met all bioavailability and content standards.

One sample out of six vitamin B-2 products did not dissolve, and one out of five samples of vitamin B-1 had the same problem. Up to five samples were tested out of each bottle of each brand.

These tests illustrate an important problem. The health food industry has been exempted from government regulation through very effective lobbying. Certain rare diseases can be treated with nutritional substances that are manufactured only by this unregulated industry, yet when a patient buys a product from a health food sore, they don't know whether the product contains too much or too little of the active ingredient, whether it will dissolve in the stomach, or whether the product actually contains an active ingredient at all.

Health food manufacturers can change their products any time they choose to do so because they are not regulated by the government. This is what happened with L-tryptophan when a new manufacturing process caused several hundred deaths and permanent crippling of innocent people who thought "natural" products are safe. A few years ago 100 babies died of vitamin E because there is no regulation of vitamins even when they are obviously used for medical purposes.

Nutritional supplements have been unregulated since the 1970's because former Senator Proxmire passed a law prohibiting FDA from regulating the industry. FDA at the time felt that some vitamins are toxic and they should not be manufactured in doses over 150% of their recommended daily allowance (RDA). The health food industry effectively campaigned by convincing consumers that the government should not impinge on their "right" to treat themselves with "natural" health foods. Since many of these consumers use health foods because they don't trust the medical establishment or can't afford prescription medicine, they swamped Congress with letters and phone calls.

Senator Orin Hatch (R-UT) , who is a health food industry food devotee, has now taken up the cause of the health food industry. Federal nutritional labeling legislation which takes effect in two years was written in a manner that could potentially make the supplement industry answerable to FDA regulations. Thus, Senator Hatch tried to put an amendment on the HHS Appropriations bill during September that temporarily exempts the industry from regulation until Congress can hold hearings on this subject. Hatch's amendment was preceded by month's of postcards letters and phone calls solicited by health food

(contintued on p.15)

Ask the Doctor

Dear CMTA:

As an orthopedic shoe technician, I have taken a special interest in addressing gait problems. I have worked with orthopedic, neuromuscular and/or neurological ambulation difficulties. I have done some shoe modifications for a few Charcot-Marie-Tooth patients while working at an orthotics facility in Richfield, MN. It appeared that, and clients reported, some benefit was obtained by specific rocking of the sole of the shoe. The idea pursued was to move the body weight across the foot/ankle with less foot slap and, therefore, less demand on the peroneal muscles.

I have recently had inquiries from two CMT patients searching for ways to improve their gait, and I was wondering if the Association could direct me to information which could help me with these two young men and any future CMT patients.

(This question was forwarded to a physical therapist, an orthopedic surgeon and a podiatrist. Their respective answers follow.)

The Physical Therapist replies:

A "rocker bottom" shoe is a shoe whose bottom has been rounded so that the surface of the shoe which hits the ground is shaped like the rocker of a rocking chair. This allows the foot and leg to "rock" over the ground as the wearer walks. The typical use of a "rocker bottom" shoe is with people who have limited ankle movement. This rounded bottom allows the leg and foot to move over the floor as the leg would normally move over the foot. For example, an ankle cast designed to allow the wearer to continue walking has a rounded bottom or "rocker bottom." This modification allows the person to roll over the foot even when the ankle cannot move. Clogs with wooden soles have rocker bottoms and many wearers, but not all, find them easy to use.

Such shoe modifications have been helpful for some patients who have mildly weak calf muscles because the rounded surface of the bottom of the shoe makes up for the inability of the calf muscles to push the person forward. However, the rocker bottom may actually make some individuals feel even more unstable, particularly if there is considerable weakness.

The technician suggests that the "rocker bottom" has been helpful for individuals with foot slap, a tendency of the foot to slap the ground as the foot makes contact with the ground while walking. This is an inadvertent, but real effect, of the rocker bottom which can be achieved by an even easier and probably more stable modification. The rounding of the shoe bottom shortens the back of the heel which lessens the force that causes foot slap. The heel of most standard shoes can be tapered approximately 1/4 to 1/2 inch for a similar effect. Some running shoes have such tapered heels or heels which are cushioned enough to have the same result. This is one reason why many patients with CMT report such ease of walking in running shoes.

As with most things, what is good for the goose is not necessarily good for the gander. An individual considering shoe modifications should talk to a health care provider who knows the patient, the disease, and the shoes. This may be a physical therapist, podiatrist, or orthotist. Then, modifications should be undertaken slowly and their effects monitored before they are judged successful and helpful.

The Orthopedic Surgeon replies:

Rocker bottom shoes are made so that in rolling the foot between heel strike and toe-off, the metatarsal heads do not bend as much. It helps shift the weight forwards in walking and with each step, when pushing-off (in push-off), pressure is taken off the metatarsal heads which are not forced into extension. It helps people with degenerative joint disease at the metatarsal-phalangeal joints. With a cavus foot, it may help protect these joints.

With peroneal muscle and anterior tibial weakness (causing foot drop),there does not appear to be any significant benefit as rocking the sole of the shoe does not help pick up the shoe and only minimizes the "slap."

In walking, after the foot is placed on the ground (foot contact), rolling (which the rocker will do) assists in forward progression of the leg. It helps the person take a longer step.

The Podiatrist replies:

Unfortunately due to the nature of the neurogenetic process of the disease, secondary muscle imbalance is both invariable and irreversible. As a result, regardless of various combinations of bracing and physical therapy or surgical intervention, the gait will not be able to be restored to its prior pre-existing status. However, this does not preclude the opportunity to enhance a deteriorating gait pattern through a multitude of physical therapy modalities, orthotic and bracing devices as well as potential reconstructive surgical procedures. In keeping with this premise, the advent of various forms of secondary complications such as contractures, ulcerations, or premature arthritic conditions may be appropriately deterred.

Dear Doctor:

I was diagnosed at Mayo Clinic in 1975 with CMT. I seem to have a light case. My hands are weakened and I wear AFO braces. I am a 73 year old female.

My mouth has had a numb feeling for at least 5 years and my speech is partially affected. My newest problem is that I have lost most of my sense of smell gradually within the last year. As a result, food has lost nearly all of its taste.

My doctor wants to know if there is a connection to CMT. I subscribe to the newsletter, but have never read about my mouth and eating problems.

The Doctor replies:

In the letter, the 73 year old female speaks of numbness in the mouth, some impairment of speech, and lost sense of smell. In general, CMT patients do not manifest these symptoms. Loss of the sense of taste or smell in the elderly is much more common than is recognized. It is important to rule out chronic sinus infection or growths in the nose or sinuses or olfactory nerves that may affect the sense of taste and smell. The involvement of the speech mechanism and numbness in the mouth suggests involvement of systems that are again not commonly involved in CMT and suggest other diagnostic possibilities.

The most important thing would be to rule out chronic infection or other illness that may be associated with this problem. Some paraneoplastic syndromes may affect the autonomic nervous system, and this may lead secondarily to an altered sense of taste, peculiar sensations in the mouth and a dry mouth.

Patient Profiles:

CMT and Kids

Joshua, Jessica & Justine Evans

Jessica Elizabeth Evans is a bright, perky 7 year old who lives in Orlando, Florida, with her Mom, Dad, brother and sister. Jessica was diagnosed with CMT at the age of 4. She is the sixth generation in her family to be affected.

Jessica is an avid learner and her favorite hobby is reading. She loves trips to the library. She read well long before she entered school and is currently being home-schooled by her mother so that she can work at an accelerated pace. Jessica recently won the spelling bee sponsored by the home and school association at the 2nd grade level.

Jessica is very involved in the activities of her family, neighborhood and church. She participates in weekly roller skating and tumbling classes. She rides her two-wheel bicycle in the neighborhood with her 4-year old brother and 6-year old sister. She is also involved in the Christian Scouting program at her church.

Best of all, according to Jessica's mother, she is a very happy, content child who is determined to accomplish whatever goals she has before her!

Seth Drzewicki

Seth Drzewicki was born on December 12, 1980 a healthy baby, developing according to the normal schedule and walking at one year of age. It wasn't until about three that Seth's continuous stumbling started to worry his parents. Even his preschool teacher commented on his frequent stumbling and falling.(This pattern of Seth's teachers commenting on his lack of coordination would continue until Seth's diagnosis of CMT.) Seth's pediatrician was consulted and a neurological workup of skull x-rays and EEG was normal. He assured his parents that Seth was just clumsy because of his height and weight. As Seth was a full head and shoulders taller than the other 3 year olds, it was easy for them to accept this.

At 8, an orthopedic surgeon was consulted because the stumbling seemed worse, but also because the third grade children began making fun of him.This doctor could not find anything wrong either except for the fact that Seth walked on the inside of his left foot. He recommended a lift that was glued on the outside of the shoe and cost about $ 6. Visits with this doctor continued for two years until a friend of his mother's pointed out to her how badly Seth's left foot curled in and how often he stumbled and fell down. As Seth's mother says, " It may seem surprising that a mother would not notice this, but I think it was because I so desperately wanted to believe the doctors that nothing was wrong with my only son."

At age 10 Seth saw an orthopedic surgeon who had treated him for something else when he was a toddler. The doctor watched Seth walk down the hall and turned to his mother and said."Yes, Laurie, Seth does have a problem." After an examination of Seth's legs and feet, the doctor told them that Seth had peroneal atrophy. In 15 minutes, someone knew why Seth couldn't walk like other children. Later, Seth confessed that he couldn't climb fences or trees like other kids, either.

The neurologist was the person who diagnosed Seth's problem as Charcot-Marie-Tooth disease. He ordered lab work and an EMG which confirmed the diagnosis. He was helpful in explaining CMT to the family, but didn't want to see Seth again for six months.

At the University of Michigan, Seth was fitted with bilateral ankle/foot orthoses and started on a physical therapy routine. He also underwent another EMG which helped to classify his CMT as type II.

It has been slightly over two years since Seth's diagnosis. He wears his braces pretty faithfully even though he would prefer not to have them. The children in his sixth grade class have many questions about his CMT, but only a few have made jokes or poked fun at him. He participates in almost every sport available through his physical education program. He has trouble with tumbling and cannot do cartwheels or work on the

balance beam. Otherwise, his mother calls him a normal 12 year old boy: he hates homework, housework, yardwork, but loves to play outside with friends.

Seth's diagnosis was the first on either side of the family. Seth's doctors in Ann Arbor feel that his CMT is the result of a genetic mutation, but that hasn't stopped his family from keeping their eyes open to the walking characteristics of the family members.

Hearing the diagnosis of CMT was really upsetting to Seth and his parents. They don't discuss why this happened to Seth, probably because it wouldn't serve any useful purpose. They give Seth love, support and encouragement while he tries to live his life the best way a 12-year old is able.

Seth is active in softball and plays first base on a co-ed team for kids from 6th through 8th grade. He is taking golf lessons, plays pick-up basketball and is an altar server at his church.

Seth has two brothers and a sister from his father's second marriage. The children visit and get along well. Seth would like to hear from CMTA Newsletter readers age 10-14. Write Seth at: 408 Sharpe St. Essexville, MI 48732-1641.

Andrew O'Brien is 17 years old and entering his senior year at North Penn High School in Lansdale, PA. He is a bright young man, deeply involved in his high school marching band, the in-door percussion ensemble, the Astronomy Club, Engineering Mentorship and his part time job. His career path is not certain; his interests include international business, engineering and medicine.

Although he had to give up marching with his saxophone because of CMT, Andrew stayed with the band and has expanded his musical capabilities to include the tympani and various other percussion instruments along the sidelines. This new experience led to participation in the in-door percussion ensemble. Both band and the ensemble require a huge commitment in time and personal discipline; however, the results have been fantastic. The Marching Band won the Pennsylvania State Championship 1992-1993 and the Percussion Ensemble won the Keystone In-Door Drill Association Championship.

In addition to a full academic program, Andrew pursued a mentorship in aerodynamic engineering at the Naval Air Warfare Center in Warminster,PA and completed an independent project of his own creation which involves laser assisted air flow visualization over tractor trailers to gain data in order to decrease their drag coefficient. He also served as vice-president of the Astronomy Club and worked 12+ hours per week at the United Artists theater.

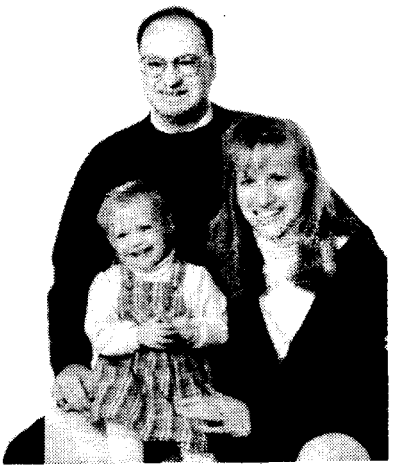
Andrew's CMT became apparent at about age three when his mother noticed he could not run or jump as easily or quickly as his preschool classmates. Actually, she believes CMT was evident in his newborn footprint which shows the right foot as curved inward when compared with the left. As Andrew's CMT progressed, the right foot has always been more severely involved. His mother and maternal aunt have CMT with symptoms that appeared in early childhood between. All have had multiple surgeries including triple arthrodeses.

Andrew O'Brien

Andrew recalls his middle school years (grades 6-7) as difficult because his CMT often set him apart from his peers. Junior high school and high school have provided many avenues for personality development, and he felt that 11th grade was a great year with many friends and activities. CMT is never too far away, though, especially when shopping for shoes. This year the challenge was prom shoes. Fortunately, Andrew got the shoes, and the GIRL on the first try!

Board of Directors Profile:

Gary Griffith

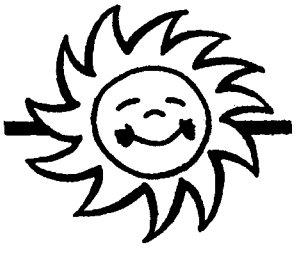


Gary Griffith & family

One of the newest members of the CMTA's Board of Directors is Gary Griffith. He actively devotes time to raising money for charity and is a lawyer who specializes in banking law, municipal corporate law, and hospital and health care law. His experiences in the legal field and in charitable fund raising make him a valuable addition to the Board.

Gary is married to Sharon Griffith and is the father of a two-year old daughter named Sam. He is a resident of Ocean City, New Jersey, and, in addition to his legal work in Atlantic City, he owns the Harbor Light Christian Book and Gift Shop in Ocean City and is an instructor at Atlantic Community College where he has been on staff since 1980.

Gary's civic, charitable, and professional commitments are many, but he has most recently been elected to serve a three year term on the Ocean City Board of Education.



Letters to the Editor

Dear CMTA Report Editor:

I've just returned from the Wilmington conference and want to thank the CMTA for putting it on. Just to see 140 people with variations of my walking and hands like mine helped resolve 41 years of feeling so isolated and alone. That in itself was worth the eight hour trip. I am glad that the last neurologist I saw a couple of years ago gave me the name and address of the CMTA.

I was glad to hear that children are being diagnosed much earlier today than when I was growing up. I hope that the lady from Washington D.C. was correct when she announced to the group that we were the last generation of people with CMT to grow up without knowing what was wrong.

I remember as a child trying to do what other kids did and not understanding why I couldn't. In the 5th grade, President Kennedy came out with his physical fitness crusade. In theory I thought that it was a good idea but was scared because I knew I was weak. Of course, I failed each one of the tests miserably and felt awful. My self-esteem was zero.

I was advised to lift weights and exercise but somehow deep down I knew there was more to it than that. In gym I was always last. I remember in high school the gym teacher telling me to push with my toes when I ran and that would help. I thought he was crazy. I remember thinking, "People don't have muscles in their toes, they're just these limp things on the end of your foot."

I did well at music. I took up the double bass and was first chair in the Virginia All State Orchestra for three years. Although I am very proud of that, CMT eventually got in the way and prohibited me from continuing to play. I once remember my teacher asking, "Why do you hold that bow like a death grip? Loosen Up." Well if I loosen up, the bow fell out of my hand since I really didn't have all the necessary muscles.

It seems to me that we should try to get the word out to others in the medical profession about CMT other than neurologists. For instance when in college around 1972 I was fitted for foot supports by a podiatrist. While they helped, in retrospect, I'm surprised that the shape of my foot didn't get him to suggest that I might have more than just high arches.

Finally, in my 30's I mentioned to my nurse practitioner that when I was on stage playing bass guitar I would start to lose my balance but regain it quickly before I actually fell. She got me to a neurologist who told me I had Roussy Levy (Roussy Levy is CMT with tremor) and sent me on my way since there was no cure.

I've seen more neurologists since and finally have learned a lot about CMT. I also have finally accepted it and would like to get involved with people in the Massachusetts area. By "finally accepting it" I mean that I accept the limits CMT will impose on my life but I won't let it get me down, nor will I transfer the inability to do certain things to other areas of my life that CMT does not affect. Whereas before I would feel that since I was weak, I must not be a "real man"... you know the 99 pound weakling syndrome.

So keep up the good work. I look forward to receiving my newsletter and if there is anything I can do in the Massachusetts area to help, please let me know.

J.L. MA

Dear CMTA,

At the CMTA conference in Wilmington, DE, a request was put forward for volunteers as Support Group organizers and/or contact people. I would like to offer my services as the latter.

I have what appears to me a "mild" case of our favorite malady, when compared to some of the friends I have met at the two meetings I have attended. At 45, my legs are to the point of developing a slight foot drop, while my hands are just now showing signs of weakening. I am still mobile without the use of walking aids and am still able to operate a rather large motorcycle (much to the chagrin of my wife.)

I think that just talking to someone else who shares this genetic "oops" may be beneficial. Like many CMT people, I was not aware of the exact nature of my problem until I was 35 years old. Up to that time I thought it was a "mild case of polio". Sound familiar? After putting a name to my problem, the next thing I needed was to meet other people who had a proper gait! The CMTA solved this problem quite well. Thank you again for your efforts on our behalf!

R.K. Baltimore, MD

Dear CMTA,

Thank you so much for your speedy reply to my letter regarding my son.

Based upon your suggestion, I did contact the Springfield Rotary Club. The Director was wonderful! After explaining the situation and also that this was not a "hardship" case as both my husband and I are gainfully employed, her first words were that the Rotary was there to help, regardless of "employment status"; that my son did qualify for their assistance; and that I should never deny my son anything that he was eligible to receive! For a period of one year, they will purchase every other pair of shoes. Each year, they will reassess his situation. Please share your suggestion with other CMT families - you, the CMTA, the Rotary, and the MDA are life savers!

On a visit to Jeremy's surgeon in late January, the doctor informed us that he has personally never seen anyone as young as our son with as severe involvement of the feet. Even after the tendon transfer on his left leg and foot, the foot still "flips." Although the surgery wasn't as successful as we had hoped, our son says that his foot does not hurt now and he really wants to have the other side done.

I'd like to tell you, too, that the CMTA Report has really been helpful; not just to me, but my son also reads it and I also share it with his grandparents. They have gained better insight into their "special" grandchild! I hope that someday there will be a conference close and I can personally thank you for your support.

V.B. Springfield, OH

Dear CMTA,

I appreciate your newsletter and all the helpful information and updates that it provides. Thank you for all your efforts.

Can I tell you about myself? I was diagnosed by default - my daughter having been diagnosed about ten years ago at the University of Miami. At various times we were without insurance and she was assisted and examined at the MDA clinic. She has orthotics for her shoes,keeps her weight down, exercised regularly and works out with weights, so she is managing with her disability very well. Her feet are very deformed and surgery may be necessary in the future.

(continued on p. 11)

Letters (continued from p. 10)

I am not greatly affected, although I have never had any physical ability or strength. The only athletic activity I could do fairly well was swimming which I still enjoy. I have had many spills throughout the years and have attributed this to clumsiness and physical weakness. It was a mixed blessing having a name for my "problem." Since my daughter was diagnosed, several members of my family have been diagnosed. (I have nine living brothers and sisters.) They seem to be affected from mild to severe. I do notice these last few years (I am 51 years old) a definite loss of strength in my hands, especially the thumb area.

I would strongly caution all CMT people to recognize their disability, great or small, and know your limits; that is to say, do not push yourself. If you are feeling slightly out of it, from a cold or headache, do not do those tasks that can wait until tomorrow. This is the most important advice I can impart. It comes from personal experience. I wasn't feeling well and instead of taking it easy, I behaved like a "normal" person, pushing myself to complete my tasks. I fell and broke my ankle. Surgery with plate and pins was required. I had good care and was delighted with the therapy. I am completely recovered;however, I did lose my job and couldn't find another one for almost a year. I am back to work now for the past three months and enjoy walking and swimmming.

Please ask CMT people to know their limits - if you are tired or not feeling 100% - don't push. You are putting yourself at risk. A broken bone is a terrible trauma, even more so if you have CMT. You must be ever vigilant to avoid setting yourself up for an accident.

K.V. Boynton Beach, FL

Dear CMTA,

Thank you for making those informative tapes. I finally had a chance to listen to/watch the Physical Therapy one. That plus the timely article by Ann Beyer in the Spring CMTA REPORT was enough to get me to call Dr.Lovelace and ask him to refer me to the MDA clinic at Columbia-Presbyterian for an evaluation. Before CMT surfaced, I used to work out with weights, but I couldn't do it anymore. Nevertheless, I want to know which muscles are weak and see what I can do to improve them.

I'm 48 and starting work on a Master's Degree in teaching English as a Second Language. It's physically challenging for me to take the subway to class and teaching jobs, but well worth it to me because I love what I'm going.

Recently, I finished writing to all the doctors who misdiagnosed me or didn't even try, in the hope that they will learn about CMT and be able to recognize it. (19 of them) I called the nicest doctor and he was sorry to hear that I have CMT, but thankful I called to tell him so he could learn from me. He said I made his day.

Thanks to the CMTA I don't feel so alone with this problem. Thanks for being there. If you find any good information about Type V, please let me know or publish it if you can. There aren't many of us, but our central nervous systems are very affected.

J.R. Brooklyn, NY

Dear Friends,

I was referred to you via the UCLA Medical Center Library in Los Angelges. I'm so grateful to have found that there's an organization to help me find out just what it is that I have.

A few years ago, I injured my back while working on a restoration job on an old piano. This year I finally got tired of throwing money at chiropractic adjustments, and decided that I would go "the medical route." The neurosurgeon who is about to operate on my spine referred me to a neurologist for an EMG as part of my workup for surgery. The neurologist discovered that I had no response in my right peroneal nerve, and obvious atrophy and weakness in my feet as well as sensory loss in one foot. Later, he told me that I have Charcot-Marie-Tooth type II disease; however, his experience with this is so limited that he couldn't answer my questions except to say that it wouldn't be a bad idea to get disability insurance.

On my own, I've read whatever articles I could find in the local hospital medical library about CMT, and as a result, I've gotten some answers to numerous things that have been nagging me for years; for example, I thought that the weakness in my legs was due to "slowing down" at age forty. I used to be somewhat well known as a fast worker as a piano rebuilder, but in the past few years, my body simply will not do the things it used to do. I've always been extremely strong and fit, and I just couldn't explain what was happening to the endurance in my legs. Also, I've had fasciculations in my arms and legs for years, as well as tingling and "humming" sensations in my feet and lower legs which I attributed to the back injury and possibly my coffee drinking.

Please forgive the rambling narrative about all of this, but I'm anxious and hungry for information. My doctors all seem to have no idea of what to expect, or how long I'll be able to work; in fact, they almost seem uncomfortable with the subject of neuropathies in general and CMT in particular. Is this a common experience among folks with this disease? If you have any reading materials or referrals, I'd be interested.

So glad to have found you; thanks for taking the time to listen.

R.M. Yucca Valley, CA

Dear Staff,

Thank you so much for the information you sent me regarding CMT. I learned more from those two booklets than I have in a whole year of dragging my son from doctor to doctor.

You indicated that the next conference would be at the Crystal Clinic in Akron, which is practically in my own back yard. I hope to attend. I also hope to be able to consult with Dr. Alexander for a second opinion as to the best course of treatment for my son.

Thank you again.

K.L. Hinckley, OH

Dear Staff,

Back in the beginning of March, shortly after my daughter was diagnosed with CMT, you spoke with me at length explaining to me about CMT and addressing many of my concerns. I appreciated all the patience and assistance you gave me in understanding what my daughter most likely faces with this diagnosis.

Your words were very encouraging and hopeful and I felt alot better after our conversation. Since then, my husband has been diagnosed with CMT as well and now we have even more questions.

We are interested in joining the CMTA(enclosed is a check) and receiving information about the CMTA conference to be held in San Diego.

Thanks again for your support.

E.C. Newark, CA

Support Group Notes

A primary goal of the CMTA is to become a truly successful advocate for those with CMT. Its message must reach the patients, their families, and the medical and research communities. Patient family support groups help carry out this function.

There are many CMTA support groups, but more groups are needed. The CMTA will help you set up a group in your area. For information about forming a group or being a local contact person please inform the CMTA by mail or call 215-499-7486.

Perhaps there is a group meeting near you. You are cordially invited to join these groups in their upcoming events.

Alabama - Greater Tennessee Valley
Bill Porter 205-386-6579W;205-767-4181
Meets at ECM Hospital, Florence, AL

California– Los Angeles Area
Oxnard-Thousand Oaks
Janice Hagadorn (805) 985-7332

Adelanta (High Desert)
Mary'L Michels (619) 246-7807

Canyon Country - Saugus
Sheila Levitch (805) 254-5322
Denise Miller (805) 251-44537

California– San Diego
Gary Oleze (619) 944-0550

California– San Francisco
David Berger (415)491-4801

California-Santa Rosa
Freda K. Brown (707)573-0181

Colorado– Denver Area
Dr. Gregory Stilwell (719) 594-9920

Florida- South
Robyn Cohen (407) 622-5829

Massachusetts– Boston
Donald Hay (617) 444-1627

Massachusettes- Southboro
Jim Lawrence (contact person)
(508) 460-6928

Michigan– Brooklyn
Robert D. Allard (517) 592-5351

Michigan– Detroit
Suzanne Tarpinian (313) 883-1123

Mississippi- Jackson
Julia Prevost (601) 885-6482
Henry & Brenda Herren (601) 885-6503

Missouri– Kansas City
Sandra Toland (816) 756-2020

New Hampshire - Southern
Mary Nightly (contact person)
(603) 598-5451

New Jersey– Central
Janet Saleh (908) 281-6289
Somerset Medical Center
Sommerville, NJ 08876

New Jersey– Northern
Teresa Daino (201) 934-6241
Meetings: Englewood Hospital
Clinic Conference Room
350 Engle Street, Englewood, NJ

New Jersey-Millville Area
Linda Muhlig (contact person)
(609)327-4392

New Mexico
Jesse Hostetler (contact person)
(505) 536-2890

New York - Brooklyn
Alan Latman (contact person)
(800) 227-1343

New York City
Diana Eline (201) 861-0425

New York– Long Island
Lauren Ugell (516) 433-5116

New York– Rochester
Neale Bachmann (716) 554-6644
Bernice Roll (716) 584-3585

New York– Westchester County
Kay Flynn (914) 793-4710

North Carolina–Eastern
Susan Salzberg (919) 967-3118
(919)286-0411 (x6586) days
Durham VA Medical Center

Ohio– Cleveland
Norma Markowitz (216) 247-8785

Pennsylvania– Delaware Valley
Dennis Devlin (215) 269-2600 work
(215) 566-1882 home

Pennsylvania-Duryea
Patricia Zelenowski (contact person)
(717) 457-7067

Texas– Greater Dallas Area
Dr. Karen Edelson, D.P.M. (214)542-0048

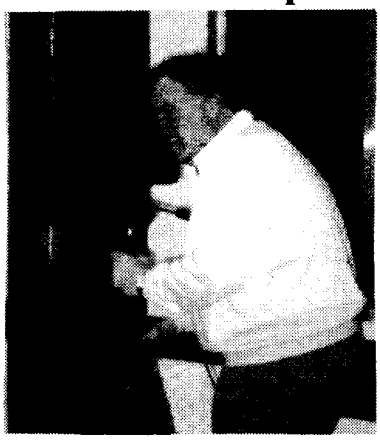
Utah– Salt Lake City
Marlene Russell (801) 966-7563 home
(801) 565-1212 work

Virginia– Tidewater Area
Mary Jane King (804) 591-0516
Thelma Terry (804) 838-3279

West Virginia- Central
Joan Plant (304) 636-7152 (after 6pm)

Washington, DC - Baltimore, MD
Lorraine Middleton (202) 362-4617
Robert Kight (contact person)
(410) 668-3054

support group corner
support group leader profile



Dennis Devlin is the new support group leader in the Southeastern Pennsylvania region. As such, he has the fortune (or lack thereof) to become a dependable volunteer to the office staff at the CMTA. Dennis is most visible in his position as semi-official video-taper at regional conferences and support group meetings. (See picture.)

Dennis lives in Media, PA and is an accountant at an area hospital. He has been married for 22 years to his wife Sue and they are the parents of Kathy (20), Jim (18) and Dan (16). His oldest and youngest children have also been diagnosed with CMT.

Dennis diagnosed at 14, had a triple arthrodesis and toe straightening at 16. He never knew that his problem was named CMT until his children were diagnosed. Ten years ago, he stopped wearing his braces and currently gets by with a pair of dress boots which he wears for business. At Christmas time, Dennis has an alter ego as he becomes Santa Claus for his neighborhood, his church, and various charitable and commercial enterprises in the Media area.

His main hobbies include computers, fishing, tropical fish, and spectator sports, especially those involving his children. "I watch my kids play as often as I can," said Dennis. "I don't let CMT slow me down and I try to do anything I want to, but I know my limitations and when I get tired, I rest rather than overdoing it."

Dennis is looking forward to the Delaware Valley Support Group meeting at Riddle Hospital. Give him a call and plan to attend a meeting if you live in the SE PA, Northern DE or Southern NJ areas.

Conferences Conferences Conferences

Three CMT patient/family conferences are scheduled for the Fall of 1993. The first of these conferences will be Saturday, October 2, 1993 in Houston, TX, at Baylor College of Medicine. The host at that conference is Dr. Stanley Appel, chairman of the Department of Neurology. Among the speakers will be Dr. James Killian, research neurologist and clinician and Dr. James Lupski, noted CMT researcher.

The second conference will be Saturday, October 30 at the Crystal Clinic in Akron, OH. The conference organizer is Dr. Ian Alexander, noted orthopedic surgeon. He is assembling area CMT specialists for this all-day conference.

The third conference will be on Saturday, November 6, 1993, at Children's Hospital in San Diego, CA. Dr. Paul Schultz, our host neurologist, has arranged a full day of presentations by area CMT experts.

Each conference will be an all day program of information about CMT from neurologists, orthopedic surgeons, physical therapists, geneticists and other experts on CMT. Lunch is included in the registration fee for each conference. All speakers include time in their presentations for questions from the audience. Each conference begins with registration at 9:15AM and concludes about 4:00PM. The all-inclusive cost for each conference is $20.00. Directions to the conference will be sent to you upon receipt of your reservation.

Fill out the registration form on this page and return to the office of the CMTA by the date specified on the order form. **Please mark carefully which conference you are registering for.**

Houston / Akron / San Diego CMT Conference Registration

Name(s) of attendees _____

_____

Address _____

_____

Telephone Number _____

Fee for one person $ 20.00

(Fees include morning coffee, lunch, and all postage and mailings.)

☐ **We are attending the AKRON conference**

☐ **We are attending the HOUSTON conference**

☐ **We are attending the SAN DIEGO confernce**

Number of persons attending _____

Amount of money enclosed $_____

Return this form by September 24, 1993 for the Houston conference
October 22, 1993 for the Akron conference
November 1, 1993 for the San Diego conference

To: **Pat Dreibelbis**
CMTA
601 Upland Ave.
Upland, PA 19015

Delaware conference participants

Delaware Conference Tapes Available

The VCR tapes from the April 1993 CMT patient/family conference are now available. The day's activities have been condensed to two 120 minute tapes. Topics covered at the conference were Adult and Pediatric Neurology, CMT Orthopedic Problems both Foot and Hand, Physical Therapy, Genetics, and Psychological Considerations. The speakers were all experts in his/her field and were very well received by the audience of over 140 attendees. The cost of the tapes is $25.00 per set of two tapes. To order, complete the order form on page 15 of the newsletter.

Health Insurance for People With Genetic Disorders:
A Report from the Human Genome Project

Editor's Note: The following report was released in May 1993 from a group studying genetic information and its affect on health insurance. The study was reported to the Ethical, Legal and Social Implications Branch (ELSI) of the Human Genome Research Project. This is a timely and critical issue for everyone and becomes particularly relevant for the CMT community because of the intensive genetic research on CMT disorders.

A task force created to assist the impact of emerging genetic information on health insurance coverage presented its report to the Joint NIH-DOE working group on the Ethical, Legal, and Social Implications (ELSI) of Human Genome Research at the Working Group's regular meeting May 10, 1993. Among its seven recommendation, the report concludes that genetic information should not be used to deny health insurance to anyone and that health insurers should consider a moratorium on the use of genetic tests in underwriting.

An important outcome of modern genetics research will be a vast increase in the kind and amount of genetic information available about individuals. Genetic information in the form of genetic tests can help predict a person's risk of disease and may alert that individual to take measures to prevent or lessen the consequences of the disease. "At the same time," the report states, "such predictive genetic information could...enter into decisions whether to seek (on the part of individuals) and whether to sell (on the part of insurers) health, disability, and life insurance."

The ELSI Working Group formed the Task Force on Genetic Information and Insurance in May 1991 to define and offer solutions to the dilemmas surrounding the potential health benefits of genetic information and the potential harm from using that same information to deny health insurance. Health insurance, usually through employers, is the main mechanism in the U.S. whereby people gain access to health care.

Headed by Dr. Thomas Murphy, of Case Western Reserve University, in Cleveland, the Task Force consists of representatives from the Working Group as well as from the health insurance industry, genetic disease organizations, health policy scholars, and genetic services providers.

Until recently, information about an individuals's risk of disease was obtained primarily from the family medical history; genetic testing options were limited and detected only a small number of relatively rare diseases. But in the past 5 years, as genetics research has accelerated, more that 50 new tests for genetic conditions have been developed. Tests for susceptibility to common diseases, such as cancer and heart disease, are currently being developed. Because the amount and kind of genetic information available to insurers is likely to increase, "barriers to adequate health care coverage will grow for a substantial number of Americans," the report says.

In the report, both insurance companies and consumers express concern over the potential for misuse of genetic information. People might decline genetic tests that could help detect early, treat, or even prevent disease if they believe insurers, who have access to their medical records, could use the test results to reduce or deny coverage. Insurers are concerned that people who learn of a susceptibility to a disease by way of genetic tests will purchase additional coverage at a price that does not reflect the likely cost of claims by that individual when he or she becomes ill.

Furthermore, the report says, defining information in a medical file as genetic or non-genetic, for the purpose of disclosure to insurance companies, will become increasingly difficult as genetic research links measurable characteristics, such as high cholesterol, to gene function. And many disease are known to result from combinations of genetic and environmental influences. "It is unrealistic to believe that insisting on physical segregation of genetic from non-genetic information in the medical record would in practice keep information from underwriters. Nor would it be an effective means of assuring that people with genetic health risks have access to health care coverage."

The report makes the following recommendations, which, according to the Task Force, should be taken as a package. "If we desire a health care system that does not erect barriers to participation for people whose genes place them at increased risk for disease, then nothing short of the comprehensive changes recommended in this report is likely to present that exceedingly undesirable outcome."

In anticipation of fundamental reform in the financing and delivery of health care in the U.S., the Task Force on Genetic Information and Insurance offers the following recommendations. The recommendations concern health care coverage and should not be applied uncritically to other forms of insurance, such as life or disability income insurance.

1. Information about past, present or future health status, including genetic information, should not be used to deny health care coverage or services to anyone.
2. The U.S. health care system should ensure universal access to and participation by all in a program of basic health services that encompasses a continuum of services appropriate for the healthy to the seriously ill.
3. The program of basic health services should treat genetic services comparably to non-genetic services, and should encompass appropriate genetic counseling, testing and treatment within a program of primary, preventive and specialty health care services for individuals and families with genetic disorders and those at risk of genetic disease.
4. Insurance premium rates for individuals and families for the program of basic health services should not be affected by information, including genetic information, about an individual's past, present or future health status.
5. Participation in and access to the program of basic health services should not depend on employment.
6. Participation in and access to the program of basic health services should not be conditioned on disclosure by individuals and families of information, including genetic information, about past, present, or future health status.
7. Until participation in a program of basic health services is universal, alternative means of reducing the risk of genetic discrimination should be developed. As one step, health insurers should consider a moratorium on the use of genetic tests in underwriting. In addition, insurers could undertake vigorous educational efforts within the industry to improve the understanding of genetic information. Copies of the full report are available from: Ethical, Legal, and Social Implications Branch, National Center for Human Genome Research, BLDG. 38A, Room 617, 9000 Rockville Pike, Bethesda, MD 20892.

Dietary Supplements
(cont'd from p. 6)

magazines and stores telling people that their "right" to treat themselves was being seriously threatened by federal nutrition labeling legislation. However, the House of Representatives refused to accept the amendment, so he is trying to attach it to the "FDA USER FEE" legislation.

Sadly, patients with valid medical needs for nutritional supplements can not trust the products they buy, but ethical drug companies are unwilling to manufacture these supplements and get them approved as prescription drugs. Once a company spends the time and money to get a new drug approved, they do not want to have their market taken away by a health food manufacturer who has no research and development expenses to drive up the cost of making the product available. Insurance companies, not knowing the difference, can refuse to reimburse for a prescription product as long as they know it is sold somewhere as an over-the-counter non-prescription preparation.

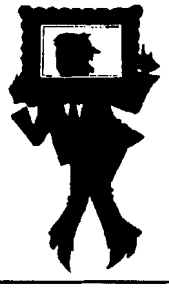
NORD intends to publicize the results of the Duke University study... When they (people) pay good money to but a product, it seems to us that they should get the labeled dosage and the product should dissolve in their stomach. FDA should regulate these products, even if they have little or no medical value for the average person.

We want to hear from YOU!

Write us at:

Letters / The CMTA
601 Upland Avenue
Upland, PA 19015
215/499-7486

Do you know of anyone famous who is a CMT patient? Do you know anyone famous who would advocate for CMT?



The CMTA is looking for a well-known person to be a spokesperson for CMT. If you know of such a person, contact the CMTA; we will do the rest.

CMTA Membership/Order Form

Name: _____

Address: _____

_____

Phone Number: _____

Tell us about yourself:

☐ CMT Patient ☐ Medical Professional

☐ Interested Supporter ☐ CMT Family Member

Enclosed is:

☐ $25 ☐ $50

☐ $100 ☐ other

for my membership in the CMTA (newsletter included in membership)

☐ At this time I cannot contribute to the CMTA but would like to receive mailings.

Publications and Tapes available from the CMTA

(Check to order)

☐ VCR Tape - CMT Neurology ($15)

☐ VCR Tape - Physical Therapy & Occupational Therapy ($15)

☐ VCR Tape - CMT Genetics ($15)

☐ VCR Tape - Orthopedic Surgery & CMT ($15)

☐ VCR Tapes (2) - Wilmington Del. Conference ($25)

☐ Handbook (16 pp.) - CMT FACTS I ($3)

☐ Handbook (24 pp.) - CMT FACTS II ($5)

☐ Transcript - San Francisco CMT Conference ($5)

☐ Letter - to Medical Professionals regarding the drug list (free to members with self addressed stamped business envelope)

☐ List - Physician Referrals (by state) (free to members with self addressed stamped business envelope) please list states:_____ _____ _____

☐ Medical Brochure - CMT (gray brochure) (one copy free with self addressed stamped business envelope)

Contributions are tax deductible.
Please make checks payable to the CMTA.

Total amount enclosed: _____

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.

MEDICAL ALERT

Certain Drugs Toxic to the Peripheral Nervous System

This is a list of neurotoxic drugs which could be harmful to the CMT patient.

Adriamycin
Alcohol
Amiodarone
Chloramphenicol
Cis-platinum
Dapsone
Diphenylhydantoin (Dilantin)
Disulfiram (Antabuse)
Glutethimide (Doriden)
Gold
Hydralazine (Apresoline)
Isoniazid (INH)
Mega Dose of Vitamin A
Mega Dose of Vitamin D
Metronidazole (Flagyl)
Nitrofurantoin (Furadantin, Macrodantin)
Nitrous Oxide (chronic repeated inhalation)
Penicillin (Large IV doses only)
Perhexiline (Pexid)
Pyridoxine (Vitamin B6)
Vincristine

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

CMT FACTS II NOW AVAILABLE

The sequel to that "best seller" CMT FACTS I is now available and appropriately named CMT FACTS II. It covers topics pertinent to the CMT patient and family which were not covered in the first booklet. Some of the articles in this new publication include the American with Disabilities Act, Orthotics, Rehabilitative Medicine, Neurotoxic Drugs and Anesthesia, and Living with a Rare Disorder. The cost of this 24 page booklet is $5.00 and may be ordered by completing the form on page 15 of this newsletter. CMT FACTS I may also be ordered and the cost for this 16 page booklet is $3.00.

CMT...

..... is the most common inherited neuropathy, affecting approximately 125,000 Americans.

..... is also known as peroneal muscular atrophy and hereditary motor sensory neuropathy.

..... is slowly progressive, causing deterioration of peripheral nerves which 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, occasional partial sight and/or hearing loss problems and scoliosis (curvature of the spine) is sometimes present.

..... 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.

..... 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 the focus of significant genetic research, bringing us closer to answering the CMT enigma.

THE CMTA REPORT

information on Charcot-Marie-Tooth disease from the

Charcot-Marie-Tooth Association
Crozer Mills Enterprise Center
601 Upland Avenue
Upland, PA 19015

Non-Profit Org.
U.S. Postage Paid
Glen Mills, PA
Permit #10

TO: