

1983-2003
CELEBRATING 20 YEARS
OF LIVING OUR MISSION

Finding
a Cure

Creating
Awareness

Improving
Quality of Life

SPRING 2003

The CMTA Report

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Our Mission:

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

Our Vision:

A world without CMT.

The North American Consortium A Personal Experience

By CHARLES HAGINS, Executive Director

In December, 1998, at the Third International Conference on CMT, a decision was made to form a Consortium in North America similar to the Consortium operating at that time in Europe.

This year, that plan came to fruition as the Inaugural Meeting of the North American Consortium was held at London, Ontario, on March 6-8, 2003, sponsored by the CMTA.

Ann Lee Beyer, Chairman/President of the CMTA, organized the event in conjunction with Dr. Michael Shy of Wayne State University and Dr. Angelika Hahn of London Health Sciences Center.

Seventy international CMT researchers presented their findings at this inaugural event. The consortium concept allows for the exchange of information and materials, stimu-

lates collaborative research, encourages the training of young scientists, and facilitates the pooling of resources. As Dr. Peter DeJonghe stated, "the consortium concept creates a deadline for results."

Although the presentations were technically beyond me, there was excitement about each topic and the subsequent discussions that followed. Another first-time attendee, Richard Sharpe, Treasurer and Secretary of the CMTA, remarked that he had never in his life learned so much about the inheritance patterns of the disorder. He, too, felt it was simply an exciting experience to be around so many dedicated and scholarly men and women who are working diligently toward solutions for the many problems associated with CMT.

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More than seventy international CMT researchers and clinicians met in London, Ontario, Canada for the inaugural session of the North American CMT Consortium.

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NORTH AMERICAN CONSORTIUM

(Continued from page 1)

Dr. Michael Shy, one of the organizers, believed the high point of the meeting was the quality of the program. "Four new genes that cause CMT were identified and presented for the first time at this meeting. Many papers presented information on the cell biology of how particular mutations cause CMT. Information about how patients' disabilities relate to cell biology were also presented. None of this information was available at the CMT meeting in Quebec in 1998. A second high point was the camaraderie of the various investigators. Many of the researchers now see each other as members of a "CMT research family" and multiple collaborations were started at this meeting which will bear fruit in the coming years. Finally, a strong relationship was established with European investigators so that an International CMT network has been set up. None of this would have been possible without the CMTA. Thanks from all of us."

Abstract presentations ranged from technical topics such as "The Regulation of the Peripheral Myelin Protein-22 Gene in Transgenic Mice" to more practical topics such as "The Effects of Resistance Training and Creatine Supplementation on Fiber Morphology and Functional Ability in CMT Disease" and "The Effects of AFOs on Gait...in CMT."

The break after each abstract presentation was greeted with more discussion and collaborative networking. It was quite common for several researchers to break away in smaller discussion groups.

On Saturday evening the CMTA sponsored a banquet dinner for the attendees. At the dinner, Ann Lee Beyer presented a Recognition Award to Dr. Peter DeJonghe, who was the Plenary Speaker and had traveled from Belgium to help advise the new North American group.

Dr. Michael Shy announced that Dr. Peter DeJonghe and his wife have agreed to coordinate next year's consortium to be held in Belgium with the idea that the event would rotate between North America and Europe every other year from now on.

With the high level of enthusiasm and commitment demonstrated by the consortium's organizers, sponsors, and researchers, the prospects for future advancements in finding a cure for CMT are more exciting than ever. ★

Professionals Working for CMT Patients: Lisa L. Baumbach-Reardon, PhD

I am pleased and honored to have this opportunity to reflect on my involvement in the CMT research community. Compared with some of the more seasoned investigators, I am still a new “kid on the block,” but I guess after serving for almost ten years on the CMT Association Medical Advisory Board I have earned a little seniority!

After I completed my postdoctoral molecular genetics training with Dr. Thomas Caskey at Baylor College of Medicine in 1989, I went to University of Colorado Health Sciences Center to complete a formal human genetics fellowship. It was in this period, that I really learned what it was like to translate research “bench” work into a clinical setting, and I was hooked! I trained in neurogenetics under the supervision of Drs. Steve Ringle and Hans Neville, to whom I am indebted for all the basic information in the fields of neurology and neuromuscular disease they provided a fledgling “neurogeneticist.” During this time, I was able to see a wide variety of inherited neurological disorders, and this training prepared me for the role I still serve today—as the geneticist for the MDA clinics at the University of Miami. The years in both my fellowships were a very exciting time—the genes for major inherited neurological disorders, such as Duchenne muscular dystrophy, myotonic dystrophy, and of course, type I and X-linked CMT were just being discovered, and DNA diagnostics for these disorders were also in their earliest stages.

It was in this atmosphere that I began my faculty position at the University of Miami in 1991. I cannot overstate the amount of excitement that was building in neurogenetics across the United States and the world from the late 1980’s to the mid-1990’s as new genes were discovered and new insights into these diseases were made at a staggering rate. It was difficult to focus on any one disease, because the flow of new information and ideas was constant. In this period, we started to appreciate not only the tremendous clinical variability of CMT

patients, and the potential for great genetic variability, but also that families with “anticipation”—disease worsening in subsequent generations—really did exist. This observation then became the basis of our first research project—investigating the molecular basis of anticipation in CMT1A families. With the help of Dr. Walter Bradley, we identified ten families with anticipation, and are still finishing our lab studies in these families. All of this work was funded by the CMTA. In the course of these studies, we detected a brand-new polymorphism (genetic change) in *pmp-22*, a gene involved in CMT1A, which was specific to African-Americans. We are just finishing our studies (which were again funded by the CMTA) of this genetic change in African-American CMT patients. Several important questions remain, the first of which concerns the origin of the mutation. If it really is restricted to African-Americans, can it be traced to any region of origin? It would be of great interest to find families of known origin or to obtain DNA from individuals



Lisa Baumbach-Reardon, Ph.D.

from various regions of Africa for polymorphism analysis. A second intriguing question concerns the possibility of correlation between occurrence of the polymorphism and disease severity. If it could somehow influence the course of CMT, might it also alter the effectiveness of therapeutics? We are trying to gather the data to begin to address questions such as these and better understand CMT1A in African-Americans, an understudied group of patients.

All of our research to date in CMT has been funded through generous support from the CMTA. I plan to continue our work in the future. There remain many interesting questions regarding the genetic basis of CMT, how different unknown modifying genes affect the clinical phenotype, and how all the cumulative information on genes and underlying nerve and cell biology can eventually lead to cures for these disorders.

(continued on page 5)

Dr. Baumbach-Reardon is an Associate Research Professor in the Departments of Pediatrics, Neurology, and Biochemistry at the University of Miami.

This survey was conducted by Anthony Redmond, the Moore Postdoctoral Fellowship recipient in 2001, and Professor Robert Ouvrier, Head, Institute for Neuromuscular Research, Children's Hospital at Westmead, Australia.

Results from the Australian CMT Health Survey

The impact of any problems associated with CMT can be highly variable, even within families. Some people show no or only minimal signs, while some have more significant disability. When people receive a new diagnosis of CMT for themselves or their children, most seek information about the various secondary features and the possible future impact. To date, the available information has been limited. The results of this survey fill in some of the gaps, provide a better idea of likely complications, and indicate how successful some of the commonly used treatments are considered to be by people with CMT.

The aims of the study were:

1. To evaluate the impact of CMT on patients' quality of life.
2. To attempt to provide better information on the range of secondary features.
3. To identify perceptions of the treatments currently used for the secondary features of CMT.
4. To identify factors that might predict the course of the disease or the outcomes of the treatment.

The survey consisted of three parts, two existing survey forms and a tailored set of questions, specific to CMT. The inclusion of existing surveys allows for comparison with other disorders for which the impacts of the illness have been evaluated.

The final response rate was 324 completed surveys received from 520 mailings (63%). The sample consisted of 132 males and 192 females. The mean age of the sample was 46.2 years, ranging from 2 to 87 years. There were 18 respondents aged less than 16 years and 61 aged 65 or older. The average age of onset was 21.2 years and the average age at diagnosis was 31.9 years. More than one quarter of people reported having a misleading initial diagnosis.

General health was evaluated using an existing instrument called the SF-36. This instrument can evaluate the impact of a disorder such as CMT on eight aspects of a person's life. The eight dimensions are:

- *Physical Function*—the person's physical ability to do things.
- *Role Physical*—the impact of any physical limitation on the person's life and relationships.
- *Bodily Pain*—the amount of pain experienced.
- *General Health*—an overall measure of the physical health of a person.

- *Vitality*—a measure of one's feeling of well being.
- *Social Function*—the impact of the disorder on the more behavioral aspects of social function.
- *Emotional Role*—the impact of the disorder on the person's ability to fulfill the emotional aspects of life and relationships.
- *Mental Health*—the degree to which the disorder leads to feelings of anxiety or depression.

The SF-36 scores range from 0 to 100, with 100 theoretically being perfect and 0 as bad as possible. Higher scores equal better health. People with CMT scored lower than the general population in physical dimensions. The physical function dimension was the worst affected, while the mental health scores remained high. The scores for men and women were very similar, but increasing age worsened the scores more than in the general population. This seems to suggest that the effects of age combined with CMT will slightly accelerate reduction in physical function throughout the lifespan. In comparison with a range of other conditions, CMT appears to lie in the mid range for impact on the eight dimensions. People with CMT reported less impairment than people with painful arthritis, and the CMT group reported broadly similar scores to people with other chronic conditions such as Parkinson's disease or stroke, after three months of convalescence.

As expected, in the foot-specific health measures, the scores for people with CMT were consistently lower than those for the general population. Within the CMT group, there was a big difference between male and female scores in relation to footwear, with women reporting more difficulty with footwear than men. The amount of foot pain experienced by people with CMT was not as bad compared with the general population, as were the scores in the other domains, such as foot function and foot health.

An overview of the most important points showed that muscle weakness was very common (reported by more than 80% of people) and was more profound in the lower limbs than the upper limbs. Severity of muscle weakness was associated with many of the features of CMT and proved to be a very useful predictor of quality of life.

Scoliosis was reported to be more common than in the general population, although none of

the other factors explored in this study predicted the severity of scoliosis. The link between scoliosis and CMT remains unclear, but fortunately only a small proportion of people with CMT suffer with significant scoliosis.

The prevalence of tremor was higher (47%) than has been reported previously in studies employing doctor-based assessment of tremor severity. This raises the question of whether patients are hypersensitive to “normal” tremor, or whether clinicians, without including patients’ perceptions, tend to trivialize minor tremor. Sensitivity to cold is often mentioned by patients with CMT, and increased sensitivity to cold in the legs and feet was reported by three quarters of respondents. Any sensitivity to cold was perceived to be worse in the legs than in the arms.

Flat feet were reported to be relatively uncommon, with fewer than 15% of respondents reporting significant flat-footedness. The high prevalence of a cavus (high arched) foot type was in agreement with the previous literature and CMT1A specifically was associated with the most high-arched foot.

Impairment of hearing and vision appeared to be reported more frequently in the CMT group than in the general population, although this study made no distinction between changes associated specifically with CMT, and other causes of hearing and vision impairment. Age was the factor most closely linked to vision and hearing deterioration. Nonetheless, the prevalence was apparently increased in the CMT respondents over the general population, and was also related to muscle weakness. This suggests some link with the physiological processes

occurring in CMT. Similarly, leg weakness also appeared to be correlated with bladder, bowel, and sexual dysfunction in people with CMT. This does appear to be an area of concern in the CMT community, and further study of this area by experts in the field is warranted.

Slowed reaction to pain was also noted, along with increased susceptibility to burns. Leg and foot weakness was again strongly related to the sensory effects reported in this section of the survey. In addition to the impairment of sensation in CMT, the survey also explored the positive sensory problems. Shooting pains in the limbs were troublesome for more than half the respondents. The prevalence of “pins and needles” is higher, affecting more than three quarters of the people with CMT. For both these presentations the lower limb is affected more than the upper limb. The other positive physical signs explored in the survey are cramps and restless legs. This survey also confirmed a high prevalence of leg cramps in people with CMT (more than three-quarters of the people) and the relationship between weakness and prevalence of cramps was again strong. Severity of leg cramps was a highly significant factor in predicting quality of life and it seems as though the importance of cramps may have been underestimated previously. Three quarters of the sample also reported restless legs, a higher proportion than reported in studies employing physician definition/diagnosis of the problem. There was a relationship between restless legs and muscle weakness.

A final report of the results will be made available when it is completed. ★

LISA BAUMBACH

(Continued from page 3)

It has been a challenge, a joy, and an honor to work in this field. I have met so many wonderful colleagues from my experience in CMT research—Tom Bird; Mike Shy; Jeff Vance; Jim Lupksi; Christine, Vince, and the scientific group from Belgium; and list goes on. I could not end without giving my thanks to some very key individuals who have helped to shape my career. First, to my lab group, who work very hard with little benefit except knowing that what they do matters. Second, I have had two tremendous mentors at my institution that have helped me in so many ways, that it would take an entire issue of the CMTA newsletter to thank them—Dr. Rodney Howell, Chairman of Pedi-

atrics, and Dr. Walter Bradley, Chairman of Neurology, whose vision and support have led to a very strong program in neurogenetics that we have built together; to my long-time friend and collaborator Dr. Phil Chance for giving me the help to get started in CMT research; and to my early supporters, including Dr. Thomas Caskey, Kurt Fischbeck, Eva Sujansky, Stephen Goodman, and others, who believed that I would one day make it as a scientist and geneticist. Last but not least, I want to thank not only the CMTA for all their support of the research in my labs over the years, but to all the CMT patients who have participated, and continue to participate in research studies—all of the knowledge that we have gained in the past 15 years is thanks to your involvement, and hopefully, will return to you very soon with real hopes for a cure for this disabling disease. ★

2ND ANNUAL “SWIM FOR THE CURE”



WHAT: 5-mile “Swim for the Cure” for CMT

WHEN: June 8, 2003

WHERE: Across the Chesapeake—*under the Bay Bridge!*

WHY: To raise money for research to help cure Charcot-Marie-Tooth disease

Encouraged by last year’s highly successful fundraising event, once again I will be swimming across the Chesapeake Bay to raise money to fund research to help cure CMT, a hereditary neuromuscular disorder that slowly cripples those affected. The CMTA is a nonprofit organization devoted to educating and serving people with CMT and their families.

Thanks for your support. —Steve O’Donnell

To read more about Steve O’Donnell’s swim, visit the CMTA’s website at www.charcot-marie-tooth.org

YES! I WOULD LIKE TO SUPPORT STEVE O’DONNELL’S “SWIM FOR THE CURE”.

NAME: _____

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Make checks payable to “CMTA” and mail to: The CMTA, 2700 Chestnut Parkway, Chester, PA 19013

The CMTA is a 501(c)(3) federally recognized charity. Your donations are tax-deductible.

You will receive a letter for your tax records indicating that no goods or services were received for your gift.

LIVING WITH CMT

Nicole Cyrille's Story

At 26, I was diagnosed with the degenerative, neuromuscular disease, Charcot-Marie-Tooth. This came as a blessing to me, after spending my entire childhood thinking I was a loser for not being able to do the things that came naturally to other kids.

When I started walking as a baby, my mother, a nurse, noticed something wasn't quite right. She took me to the doctors, who ignored her constant concerns. I remember being a small child at the doctor's office when he would check the reflexes in my knee; I had none. He didn't find this strange but told my parents to remind me to walk heel-toe because I just didn't want to do it. It was a constant tape playing through my house with my parents saying, "heel-toe Niki." In my mind I was walking right, but my body couldn't respond.

In school, my undiagnosed disability wasn't different enough to elicit sympathy from the other kids, only teasing. I endured ridicule on a daily basis because I walked funny. I wish I could say that the ridicule ended when I grew up. The only things that changed were the words adults used—she's drunk. Or my favorite—copying my horse-like walk and laughing. I cried a lot. Needless to say, the constant reprimands and teasing and the fact that I was different damaged my self-esteem.

Years later, while living in Las Vegas, I started working as a model and an actress. The work I did varied from a Bon Jovi video, to a runway show for Betsy Johnson, to other television and film productions. After meeting Mike Myers on the set of *Austin Powers*, I moved to Los Angeles to grow as an actor and study with the Groundlings.

When I noticed that my right ankle had deteriorated, I sought a doctor in Los Angeles and when he saw me walk, he asked if I had CMT. Initially, I thought he was asking if I liked Country Music Television. After he explained that it was a disease and told me the symptoms, I knew this was the answer. I phoned my mother from the examining room. She was convinced he was wrong because none of the myriad of doctors ever suggested it to her. I didn't have the tests run at that point because of my mother's insistence that it wasn't possible.

Eventually, my mother, after suffering an accident, had to undergo a battery of tests. It was then that the technician suggested she be tested

for CMT. She had it. Then I had the blood test and it confirmed what I already knew—I did too.

Career-wise, I spent a year in Nashville, where I started performing as a stand-up comic. I felt it was easier to educate people through laughter. Since then, I've moved back to LA and found the ignorance about the disease disheartening. One day I was walking around the NBC lot when an actor decided to ridicule the way I walked. It was late in the day for me, which meant my legs were far more tired and my walk was more pronounced. Another glaring moment was after telling a famous talk show host about the disease. His response was trite: with a blank expression, he asked if it meant that I couldn't go up in the space shuttle. These experiences prompted me to call the Charcot-Marie-Tooth Association and get involved.

The fact that I was willing to put myself out there as a voice for this disease led to my being named a spokesperson. My main goal is to spread awareness and compassion so that children can know early on that their physical problems don't make them any less a person. There is a sense of empowerment that comes with an early diagnosis, and I want all children to experience it. It is my passion.

Because the pharmaceutical companies can't make money from CMT, there is very little attention given to it. The current lack of a pharmaceutical protocol keeps diagnosis down and greatly hinders progress on a cure. Not to mention that certain drugs have an adverse affect on patients with CMT and have caused deaths in children that were undiagnosed until it was too late. ★

Nicole Cyrille is an actress diagnosed with CMT who has offered to be a spokesperson for the CMTA.

Robert Kleinman, Nicole Cyrille, Dick Sharpe, and Phyllis Sanders met on Long Island to plan a CMT awareness campaign.



GIFTS WERE MADE TO THE CMTA

MULTIPLY YOUR GIFT

Matching gifts are an excellent way of increasing your gift to the CMTA. Ask your employer if they offer a matching gift program. Other "power of one" examples include our golf tournaments and the swim of the Chesapeake Bay. If you have an idea for a fundraising event, call the office and we will connect you with the appropriate advisor.

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Ask the Doctor

Dear Doctor:

In a recent blood test, my internist found my homocysteine level to be too low and recommended a daily dose of a product called Isotonix. It is a powder (to be mixed with a liquid) high in Vitamin B₁₂ and folic acid.

I wondered if anyone has any information about this product and its effect, if any, on people with CMT.

The doctor replies:

I am not familiar with this brand name product. The two ingredients—B₁₂ and folate—are safe for people in general and for people with CMT, in particular.

Dear Doctor:

Do problems with CMT affect wound healing? Can you offer any suggestions on improving the healing process? I understand there is a topical product that stimulates the formation of granulation tissue. Has this been tried in folks with CMT who are having difficulty with wound healing?

Is there any change (elevation) in levels of serum muscle enzymes in people with muscle wasting secondary to neuronal pathology?

Thank you for your assistance.

The doctor replies:

A few rare kinds of inherited neuropathy cause a propensity to injury, as painful sensations are no longer perceived. Beyond that, nerve fibers are thought to help maintain the health of skin, but the molecules involved have not been identified. Only severe neuropathies that affect the innervation of skin are associated with so-called “trophic changes” of the skin, and it is possible that poorer healing could be a manifestation of these trophic changes. I do not think that most people with CMT have enough denervated skin to have poor wound healing.

I am not aware of a “topical product that stimulates the formation of granulation tissue.”

Peripheral neuropathy alone does not cause an elevation in creatine Kinase level.

Dear Doctor:

I am going to have an endoscopy and the doctor is planning to use Versed®. Is there a danger from this drug? I had the procedure previously and chose not to use this medication.

The doctor replies:

Midazolam (Versed®) is a medication used (in this instance) to assist in the sedation, reduction in anxiety, and impairment of memory in patients undergoing procedures (including endoscopies). Some medications have so-called “black box warnings,” whereby past experiences dictate caution in their use because of significant adverse reactions. Midazolam has a black box warning: “Midazolam I.V. has been associated with respiratory depression and respiratory arrest. In some cases, where this was not recognized promptly and treated effectively, death or hypoxic encephalopathy resulted. Use midazolam I.V. only in hospital or ambulatory care settings, including physicians’ offices, that provide for continuous availability of resuscitative drugs and equipment and personnel trained in their use...” This would apply to any patient. As we know, the muscles affected in CMT patients are skeletal. Overdosing of Midazolam may be a concern in CMT patients. It sounds to me that since you underwent this procedure before without midazolam, it would be safer for her to repeat what she had previously done.



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.

American Academy of Neurology Meets in Hawaii

Approximately 7,000 people attended the recent American Academy of Neurology meeting in Honolulu, Hawaii from March 29 to April 5, 2003. A number of presentations on CMT were delivered at the accredited neurology courses. Medical Advisory Board member Dr. Michael Shy discussed the diagnosis and management of patients with CMT. Another Board member, Dr. Mark Bromberg, talked about CMT as part of the larger group of peripheral neuropathies.

Other topics of interest for the CMT community were a discussion of the patterns of weakness found in CMT patients, the importance of avoiding vincristine and other neurotoxins in children with cancer and CMT, and the recent discovery of the CMT1C and GDAP1 genes. There were a total of ten abstracts on CMT and one interesting paper that discussed vocal cord and diaphragmatic paralysis in CMT due to GDAP1 mutations.

The meeting was attended by Amar Kamath, a non-voting member of the CMTA’s Board of Directors. ★



Easy Gait Brace Provides Help for Foot Drop

A previous article in The CMTA Report created interest in the Walk Right brace and this article introduces the improved bracing from the same company. It is not suitable for all CMT patients, but for those with minimal muscle loss and moderate foot drop, it is an inexpensive and less intrusive choice.

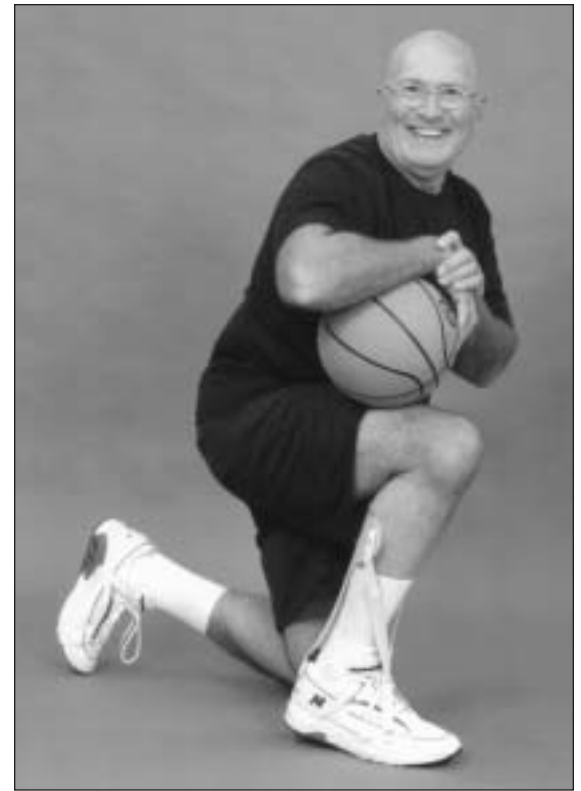
The Easy Gait brace can be snapped onto a sneaker with ease.

The Easy Gait Brace is an innovative, lightweight, custom-designed orthosis which is ideal for individuals with mild-to-moderate foot drop. The brace is cool, non-rigid, and more comfortable than heavier, more conventional AFO's. Keith Markusic, founder of Walk Right, Inc., can walk backwards, forwards, climb stairs without difficulty, and play YMCA basketball with the brace attached to his right sneaker. Dr. William Quinn, DPM, and a CMT patient himself, wears the device and recommends it to his patients who require minimal bracing. "As a surgical podiatrist with Charcot-Marie-Tooth, I wear an Easy Gait on both legs and highly recommend it for people who have some muscular function."

Compared to standard AFO bracing, the cost of an Easy Gait brace is reasonable, especially since it does not require a doctor's prescription to order and is largely reimbursable under Medicare Billing Code L1910 with a prescription. Suggested retail price is \$305 and the device can be ordered through the mail, by phone, or on the Internet at www.ezgaitbrace.com.

Some of the advantages of this brace include:

- Lightest of any AFO and drop foot brace.
- Cooler in hot weather.
- Brace can be snapped onto any lace-up shoe or sneaker.



Easy Gait braces allow a wearer to continue athletic endeavors.

- No plastic or other hard insert is needed inside your shoe.
- Increases mobility and ease in climbing stairs and inclines.
- Facilitates noticeably more even and normal walking gait. Two elastic straps provide passive resistance against the foot.
- Color-coordinated bands match your socks and shoes. Brace is unrecognizable under long pants.
- High comfort level and easy application increase length of use.
- Affordably priced for all patients.
- No doctor's prescription required. Medicare reimbursable.
- Unconditional money-back guarantee within 90 days of purchase. ★



If you have questions about the brace, call Keith Markusic at Walk Right, Inc., 1-800-467-3151 or on-line at askezgait@aol.com.

Winning...with CMT

By PEGGY SPITTLER

Encourage your child to dream and set goals. Even with CMT, he or she can still dream and achieve. Children with CMT may be unable to do some things because of their disability, but there remain many things they can do, and do well! Children need to stay focused on those things—not beat themselves up over unsuccessful attempts. Tell your children to bask in the glow of their accomplishments. Who they are has nothing to do with the way that they walk or the appearance of their hands. It's what's between their ears, and what's in their heart that's important. Encourage your child to stay focused on who he or she is and not physical appearances.

Since my mother didn't have CMT, we were never given reasons for being more tired than the other children, tripping over our turned-in feet, lacking balance, or turning our ankles and falling. My father worked every day in the coal mines without complaining. I think that being a coal miner was a good job for him

because he spent most of the day on his knees, taking the stress off his feet and legs, since the ceilings of the coal mines were too low to stand upright. Our entire family idolized our father. We thought he could do no wrong. We did not see his disability or the things he couldn't do. All we saw was a great man who worked hard, who loved his family, and loved to hunt and fish on his days off. Dad raised honey bees and spent a lot of his leisure time lying on the ground in front of the hives, watching the bees fly in and out, making honey from the surrounding fields of clover, sourwood, and buckwheat. Dad's disability did not hold him back. He lived a full rich life. He died at the age of 86 after having several strokes.

At the age of six, I started school. We had to walk to the one-room school about a mile and a half away. I have very few memories before that time. My mother was told that I had been struck with polio while visiting my grandmother the summer before starting school. I was kept home from school for the first six months. I had difficulty walking. I was held back that first year, due to the class time I missed. I enjoyed school and made good grades, but the effort it took to

Encourage your child to dream and set goals. Even with CMT, he or she can still dream and achieve.

get there and back left me exhausted. My knees had scars on top of scars due to constant falling. I was always turning my ankles and tripping over my feet.

The boys in the family who had CMT were never given limitations. I remember being warned about all the things that I couldn't do. I couldn't go swimming or ride a bike. I was always the last one picked for games or sports. I had very poor self-esteem. The shell of hurt grew harder. Every time I was called crippled or humpback, I died inside. This was an awful heavy load for such a frail little girl. I didn't want to tell my mom about all of the children

picking or poking fun at me. Perhaps, down deep I was afraid of losing her love, also. There was very little emphasis placed on the things that I could do. I always looked forward to report card time, because making good grades was one thing that I could do.

I remember going to the "Crippled Children's Clinic" with my mom and aunt. My

aunt was taking her daughter who had a birth defect that required bracing. By the age of ten, my scoliosis had become so bad that bracing was recommended. The brace was very heavy and bulky. My armpits and the front of my shoulders became blistered and sore. I began leaving the brace under the bed or in the closet. Needless to say, the brace didn't help much because wearing it was so unbearable.

In the autumn of 1959 I began high school. We were still walking a mile and a half to catch the bus that took us another six miles to high school. Changing classes with an arm full of books and participating in gym were the straws that broke the camel's back. I began having tremendous back pain and my breathing capacity began to suffer. At fifteen I was taken to the Miners Memorial Hospital in Beckley, West Virginia. By that time the curvature of my spine was almost at 75%. Surgery was recommended.

A spinal fusion was done by taking bone from my right hip to fuse eight vertebra that had been straightened by casting. The straightening process took several weeks. I can still feel the warmth of the bandages that went up around

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WINNING WITH CMT

(Continued from page 11)

the top of my head and down the other side, all the way to my knees on one side and my hips on the other. Halfway through the process, huge turnbuckles were placed along the side of the cast. As the casting continued, my body became a backward “C” that tilted toward the left. As the weeks continued, the turnbuckles were turned a little each day, until the spine was as straight as possible. After the surgery I spent about six months in the hospital. I returned home with a walking cast. I wore it for another three months. I started my junior year in the hospital with a tutor, and finished at home, still in a cast, with a homebound teacher. When the cast was removed, I was amazed at how straight my back was. It was the first time I could remember feeling good about myself. I still had a limp because my legs were still weak and my ankles still turned over, but I had a happy heart.

I began to have a social life for the first time in my life. I was visiting my sister in Alexandria, VA when I met my first husband. I had aspirations of attending college after graduation, but the lack of finances caused me to give up that dream. I got married at the age of 20. One year and one week later I gave birth to my first daughter, Coralea (Kandi).

When Kandi began walking I noticed her little feet turning inward. My heart dropped. I didn't want her to endure the heartaches of growing up as I had. I became concerned and consulted her pediatrician. He told me that she was fine and that I was worrying unnecessarily. So, I just stopped worrying. Three years and six weeks later my second daughter, Angela Renee was born. She was also a healthy and beautiful little girl. I decided to go to work so that I could

support my family. I enjoyed my job but not the long drive to get there. After my marriage fell apart, I continued working.

During one of our lay-offs I had surgery on my right ankle. Trying to care for two children after surgery was difficult, but the result of the surgery was worth it. Now I was only turning over one ankle instead of two. Around that time I decided to go to college. The financial aid that I received made it possible. My children and I lived on grants and food stamps. I had a part-time job, also. I was feeling good about my life and myself. I graduated in 1979. I worked part time at a county rehabilitation center during my last year in school and continued full time after I graduated. My present husband and I were married in August 1979. Curt had three children, two girls and a boy, so we went from a family of three to a family of seven. I went to work as a case-work aide for a home for court-ordered youth and a home for mentally retarded young adults.

I continued my employment as a casework aide until I was no longer able to work. I went on social security disability. Two or three years later I tried to return to work. I was employed as a residential program worker. I enjoyed my job and thought I could do it. After three years, I realized that my body demanded more and more rest. I decided to go on permanent disability. Shortly after that I had to have a total hip replacement. I was 51 years old at that time. Now I am 58. I have had a lot of problems with my hip replacement. It has dislocated 16 times. The doctors keep telling me that I do more than I should, but I'm afraid of becoming inactive. When I just sit around and read or watch TV, I become very weak. After a couple of days my legs feel like rubber toothpicks. But, as long as I stay busy, I do pretty well. I have to sit or lie down after working for a couple of hours. I love landscaping, gardening, and watching nature. I spend a lot of time working on church projects.

Our children are doing great; they are all grown and have children of their own. We have 14 grandchildren. Kandi and Angel both have CMT. Kandi works as a case manager in Alabama. She has three children, one of whom has CMT. Angel works part time as an accountant in her home. She has three children, two of whom have CMT. She works from the time she gets up in the morning until bedtime. She tries to lie down in the afternoon a little while before the children arrive home from school. Her legs and hands are very weak. But, she refuses to admit that she has limitations. She has them—so do I, so did my father and my grandmother, and her mother—but we are all winners because we refuse to let CMT win. ★

ATTENTION: DRUG RECALL

The Food and Drug Administration (FDA) has issued a public advisory concerning the drug phenylpropanolamine, commonly known as PPA. The drug is found in many over-the-counter (OTC) and prescription cough, cold, and decongestion remedies, as well as in OTC weight-loss products. The FDA is in the process of removing PPA from such products, and is encouraging drug companies to discontinue the sale of these items. It has been found that taking PPA increases the risk of hemorrhagic stroke (bleeding into the brain or into tissue surrounding the brain) in women, although men may also be at risk. The actual chance of having a stroke remains very low, but protect yourself and read labels carefully. If you have or are taking any products with PPA, please discontinue use immediately.

REHABILITATION AND MANAGEMENT OF CMT:

Paolo Vinci's Approach

After coping with CMT for 38 years and dealing, as a rehabilitation doctor, with about 300 people affected with CMT, I can say that CMT not only weakens the muscles in the feet, legs, and hands, and causes joint deformities and sensory impairment, but it also alters life in a global way. This is because:

- CMT generally starts in infancy and adolescence, when everybody should be allowed to live carefree and not be obliged to undergo physiotherapy 3 times a week (as happens in Italy) or wear special shoes (instead of trendy shoes) or avoid the sports and the experiences other children enjoy.
- CMT alters physical appearance in a period of life when any minimal defect is not tolerated by one's peers (The mass media places undue importance on physical perfection).
- CMT is a genetic disease and that presents ethical problems for people at reproductive age because of the risk of passing on the defective gene; this may alter the relationship with the partner and make the CMT patient feel inadequate.
- CMT impairs the most important functions of the limbs, reducing gait and grip causing efficiency at work and in daily activities.
- CMT is progressive and the patient's future may appear uncertain.

Incredibly people with milder CMT can complain more than severely affected people. Recently a woman with mild CMT complained to me that she was not able to walk as fast as her friends and could not wear miniskirts anymore since her calves had become slightly thin. She had started refusing social invitations and felt unhappy. Her major problem was not her mild neuropathy but her attitude toward the disease, because she did not know whether to consider herself normal or disabled.

A good rehabilitation approach to CMT must address, and if possible solve all the problems that prevent the CMT patient from living a full and happy life.

To address the problems briefly presented in this introduction, a team of several professionals familiar with this disease are needed: physiatrist, pedorthist, physiotherapist, orthotist, occupational therapist, orthopaedist, psychologist and geneticist.

CMT not only weakens muscles.... it alters life in a global way.

It might seem strange that I include psychologist and geneticist in the rehabilitative team, but a psychologist can be very helpful, especially with adolescents, in the essential process of disease acceptance. And the geneticist, too, is important, because knowledge of the particular gene responsible for CMT, the process of disease transmission and ways to avoid it (prenatal diagnosis, genetic pre-implantation diagnosis) permits the patient to

decide about procreation with awareness and, therefore, better his/her own future.

Of the functions directly affected by the disease, rehabilitation generally addresses stance, ambulation and handgrip. However, running, which is generally neglected, is important, if not in adults, then surely in children who cannot be forbidden to run unless one wants to make them feel different from their mates. If we cannot improve running performance, we can at least address the consequences of running on those muscles and joints.

Ambulation is generally impaired in all persons with CMT, but severity varies, depending on the extent of muscle weakness in the foot and leg, joint deformities secondary to impaired muscle strength and compensatory behavior.

In recent years, I have had the opportunity to treat only people with CMT and to dedicate many hours to each of them. This has enabled me to develop a methodology of examination that insures complete understanding of the patients' problems.

I have developed a classification scheme for patients with CMT. Patients are assigned to one of several levels of functional impairment of gait and to one of four levels of handgrip impairment. For each level there is a different rehabilitation management, consisting of proper shoes and orthotics, physiotherapy, occupa-

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This article appeared in the Spring 2003 issue of ComMent, a publication of the Charcot-Marie-Tooth United Kingdom. Paolo Vinci, MD, PhD is a practicing physiatrist in Italy.

Airline Security and Disabled Travelers

from Orphan Disease Update, a publication of NORD

People with disabilities sometimes complain that airline security procedures that were put in place after September 11 make them feel harassed. For example, blind people are separated from their guide dogs because dog harnesses set off metal detectors, diabetics must show proof of their diabetes to take hypodermic needles in their carry-on luggage, and people with artificial hips or even pacemakers are sometimes required to take their clothes off just to prove that they have scars to indicate they have an implanted medical device.



“Most ordinary passengers dread having to go through security checkpoints at airports, but people with implanted metal medical devices feel particularly hassled when they are required to prove that they are not terrorists,” said NORD’s President, Abbey Meyers. “The burden should be on security personnel to reasonably suspect illegal activity, and the burden should not be on passengers with disabilities to prove that they can’t remove metal devices that are implanted in their bodies.”

Many passengers with implanted medical devices follow the prescribed course by carrying a card illustrating the implanted device, and/or notes from their doctors. They notify the airline in advance so their disability is noted on their computer record. Yet personnel at security gates sometimes don’t believe these documents and make the passenger disrobe in a curtained booth to prove their scars.

“There is no uniformity to these searches,” says Mrs. Meyers. “Some airports are worse than others, but you never know. It is obvious that some of the security personnel have not been properly trained,” she said, noting that there are many complaints from people carrying medical equipment such as breathing machines. Regulations from the Department of Transportation say that if a person carries medical equipment, it shall not be counted as a third piece of carry-on luggage. Thus, passengers are allowed to carry medical equipment on an airplane even if they are carrying the two permitted pieces of carry-on luggage (e.g., a briefcase and computer).

NORD suggests that you download the Department of Transportation’s regulations for people with disabilities (http://www.tsa.dot.gov/trav_consumers/tips_disabilities.shtml) and carry a few pages with you so you can educate security personnel who don’t know the rules. Always ask them to call an airline supervisor if you have a problem because the supervisors are usually better trained than security personnel, who are contractors. ★

PAOLO VINCI’S APPROACH

(Continued from page 13)

tional therapy, surgery, and, last but not least, rules of life.

The first step of examination is manual muscle testing. This should be done once a year, to monitor disease progression, in contrast to the development of joint deformities and postural abnormalities perform a complete joint range-of-motion testing, and special functional evaluations, to ascertain the severity of the impairments and reduce the risk of falls. Next I try to correct supination and equinus deformity using wedges that, if they look effective, are incorporated into the soles of the shoes or foot orthoses.

I then measure the angle between the axis of each leg and the sole of the foot while the patient elevates the foot as much as possible. This evaluation is important to decide if a dorsiflexion-assisting device is necessary. For people with severe deficit of ankle dorsiflexion, I use ready-made or custom-made boots or the traditional ankle-foot-orthoses (AFOs).

As for physiotherapy, I distinguish between therapy necessary to reach a well-defined short term goal and long-term therapy. Short term therapy is very helpful. After fitting a patient with a dorsiflexion-assisting device (AFOs or drop-foot boots), short term physiotherapy is necessary to help recover physiologic range of motion at the hips, correct postural abnormalities and provide ambulation training to modify the pattern of gait and improve balance.

As for long-term physiotherapy, I do not prescribe it because it is useless and only reinforces the disease sensation. The exception is in children, who should be checked regularly and for a long time by a physiotherapist, after providing them with foot orthoses or shoes with lateral wedges, to ensure that persistent activation of the supinator muscles does not damage the knee. To adults, I say that living a life as normally as possible is the best physiotherapy. ★

“Rehabilitation management of Charcot-Marie-Tooth disease” can be ordered online from the website: www.aicmt.org/books.htm

JOINT EFFORT

Ann Lee Beyer, Chairman and President, and Richard Sharpe, Treasurer/Secretary met with Susan Wheeler of CMT...Today and their Board of Directors to discuss a collaborative venture between the two organizations. This collaboration could include dual memberships and access to each other’s newsletters for members of the two groups. A follow-up meeting will take place in May.

Chronic Pain and the Family

By **LESLIE F. MARTEL, PhD**, from the Newsletter of the Chronic Pain Outreach Association

We are born within a social context, we live within a social context, and we get ill within a social context. As self-evident as this might seem, our medical system, to some extent, tends to ignore this fact. Western medicine remains fascinated with technologically advanced methods to treat pain and, as a result, attention to the interplay between chronic pain and the family system is not studied with the same intensity. This is not to negate the importance of the medical advances but rather to emphasize that a truly comprehensive approach to chronic pain is necessary to maximize improvements for as many people as possible.

For the person experiencing chronic pain, the temptation to repeatedly involve oneself in medical procedures may be very seductive. Given a heightened level of distress in the patient, the physician or other health care provider may also experience this temptation. Both patient and doctor share a common goal—to decrease suffering. Given this, it may be helpful for doctor and patient alike to better understand how pain and the family system interact.

When first approaching a persistent pain problem, it is usual to seek physiological or anatomical causes and cures. Yet, for many individuals with chronic pain, physiological and anatomical causes provide an inadequate explanation to account for the subjective experience of suffering and for the lack of success of conventional treatments. After a comprehensive medical evaluation and an adequate trial of conventional treatment, there comes a time when both doctor and patient must acknowledge that the continued search for the “lesion” is not a productive course of action. Knowing when to “hold on” and when to “let go” is not an easy decision in any area of one’s life, and this is certainly true for the treatment of chronic pain. Once the patient and family acknowledge that the pain will not be entirely removed by medical means, the healing process can truly begin.

Physicians and health care providers are accustomed to a “systems approach” to biologi-

cal organisms. For example, it is clearly understood that a problem in the heart, lungs, and blood vessels has implications for the cardiovascular system as a whole. However, they are generally not accustomed to viewing social groups such as families in systems terms. Yet, it is just such a framework that is necessary to fully understand the complex nature of chronic pain.

There are five core family systems principles (adapted from Doherty & Baird, 1983) that are helpful in understanding this perspective:

1. The family is more than a collection of individuals. That is, the whole is greater than the sum of its parts. One must view the family as an entity in its own right with its own “life.” Knowing all of the members individually is not enough. One must understand that change or stress affecting one member of the family affects the whole family.

2. Families have repetitive interaction patterns that regulate member’s behavior. These are the implicit rules for daily living. These may be “rituals” and are often not articulated. These patterns make family life predictable and generally make life easier. On a less benign level, the patterns may “freeze” a particular way of behaving, making change difficult.

3. Individuals’ symptoms may have a function within the family. For many reasons, a symptom may become incorporated into the family interaction pattern in such a way that it seems essential for the family’s harmony and regularity.

4. The ability to adapt to change is the hallmark of healthy family functioning. Change is the ever-present challenge to families. In addition to normal life cycle transitions, illness may challenge a family’s ability to adapt to new circumstances. With illness comes the reshuffling of roles. Perhaps the primary wage earner must now become the recipient of disability payments. The ability to handle these changes with flexibility, creativity, and determination reveals a great deal about family functioning.

**Both doctor
and patient share
a common goal—
to decrease
suffering.**

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CHRONIC PAIN

(Continued from page 15)

5. There are no victims and victimizers in families: family members share joint responsibility for their problems. Family members are both actors and reactors, especially in maintaining chronic problems. There are no family villains. Like a dance, family members move with one another in ways that lead to healthful or hurtful consequences. Family systems theory encourages one to look at the interconnectedness of behavior and seek solutions in accordance with this viewpoint.

The person with chronic pain is not the only person whose life changes. Each member of the family must make adjustments which may be psychological, social, economic, and physical in nature. All involved will have thoughts and feelings about these changes and it will be essential to remove barriers in order to freely discuss these issues. Honest, straight-forward, and routine communication is the key to maintaining healthy family functioning.

The founder of general systems theory, Ludwig Von Bertalanffy, noted that “system sickness is system rigidity.” It is essential that

the family examine the patterns that have developed as a result of one of their members being in long-standing chronic pain. Has the family become “frozen” in their roles? Is there room for change that would improve the quality of life for all concerned?

When a member of the family system develops chronic pain and it becomes clear that the problem is, indeed, chronic, the family will usually reorganize in a way that allows it to keep functioning as a family unit. Once the reorganization of roles and the shift in power have occurred, there is a distinct tendency for this new order to be maintained. For example, Mr. X, who was responsible for working 40 hours/week, taking care of household maintenance, and paying household bills was relieved of these responsibilities during a long and difficult bout with chronic pain. He may be unaware that he has now adapted to a life free of these responsibilities. When he attempts to handle bills, he finds that, for some reason, he experiences more pain. His wife, who is quite caring and solicitous of him, assures him that she can handle these tasks.

Over time, a symptom may take on a life all its own. The family needs to be aware that this can occur and make a special effort to routinely assess family roles/tasks and make changes that will facilitate continued growth of the individual and family unit. By understanding that significant change is usually accompanied by some resistance to it, difficulties may be anticipated and eventually overcome.

These two points are closely related to the three previous ones. Change is an ongoing and necessary process. The ability to adapt to changes and to acknowledge what you can change and what you cannot change may potentially lessen the feeling that one has lost control of one’s life. Viable choices usually exist. Each choice has its pros and cons, but it is making choices that prevents a person or family from feeling victimized.

In essence, if chronic pain affects a given individual, it becomes essential for the health care professional, the “sufferer,” and the family to understand that the pain also affects the family. By understanding this, all concerned can view the problem in a broader context and address the issues accordingly. With increased stress in the family system, communication tends to break down, patterns become more rigid, and overall family functioning diminishes. Addressing the concerns of all those involved in a timely manner will go a long way toward identifying and being able to remedy any potential problems. ★

Help Perpetuate the CMTA’s Work

Remember the CMTA in your will. You can give hope to thousands of CMT patients by extending your support of the CMTA’s programs beyond your lifetime. Whether your legacy is small or large, you can support our programs of education, service and research by remembering the CMTA in your will.

To make a bequest of cash or other property to the CMTA, your will (or supplemental codicil, if you do not wish to write a new will) should state:

“I give and bequeath to the Charcot-Marie-Tooth Association, a not-for-profit corporation, organized under the laws of the Commonwealth of Pennsylvania, and having its principle office at 2700 Chestnut Parkway, Chester, PA 19013, the sum of \$() or () percent of the rest, residue and remainder of my estate to be used for the (general purposes) or (research fund) of the Organization.”

A bequest to the CMTA is fully deductible for estate tax purposes. Additionally, you will be providing hope to CMT patients and family now and in the future. You may wish to learn about other gift giving opportunities by consulting your attorney, accountant, and/or tax or estate planner. ★



VITAMINS: A BRIEF OVERVIEW

Vitamins and minerals are fundamental to human health, growth, and, in some cases, disease prevention. Most are consumed in your diet (exceptions being vitamins K and D, which are produced by intestinal bacteria and sunlight on the skin, respectively). Each vitamin and mineral plays a different role in health. The following outlines essential vitamins:

- VITAMIN A is important to the health of your eyes, hair, bones, and skin; sources of vitamin A include foods such as eggs, carrots, and cantaloupe.
- VITAMIN B1, also known as thiamine, is important for your nervous system and energy production; food sources for thiamine include meat, peas, fortified cereals, bread, and whole grains.
- VITAMIN B2, also known as riboflavin, is important for your nervous system and muscles, but is also involved in the release of proteins from nutrients; food sources for riboflavin include dairy products, leafy vegetables, meat, and eggs.
- VITAMIN B3, also known as niacin, is important for healthy skin and helps the body use energy; food sources for niacin include peas, peanuts, fish, and whole grains.
- VITAMIN B6, also known as pyridoxine, is important for the regulation of cells in the nervous system and is vital for blood formation; food sources for pyridoxine include bananas, whole grains, meat, and fish.
- VITAMIN B12 is vital for a healthy nervous system and for the growth of red blood cells in bone marrow; food sources for vitamin B12 include yeast, milk, fish, eggs, and meat.
- VITAMIN C allows the body's immune system to fight various diseases, strengthens body tissue, and improves the body's use of iron; food sources for vitamin C include a wide variety of fruits and vegetables.
- VITAMIN D helps the body absorb calcium which strengthens bones and teeth; food sources for vitamin D include oily fish and dairy products.
- VITAMIN E can help protect certain organs and tissues from various degenerative dis-

eases; food sources for vitamin E include margarine, vegetables, eggs, and fish.

- VITAMIN K is essential for bone formation and blood clotting; common food sources for vitamin K include leafy green vegetables.
- FOLIC ACID maintains healthy cells and blood and, when taken by a pregnant woman, can prevent her fetus from developing neural tube defects; food sources for folic acid include nuts, fortified breads, leafy green vegetables, and whole grains.

It should be noted that one can overdose on certain vitamins which become toxic if consumed in excess (e.g., vitamins A, D, E and K). For people with CMT, an excess of B₆ can be toxic to the peripheral nervous system. ★

CMTA PRESENTS AWARD TO DR. PETER DEJONGHE



Ann Lee Beyer, Chairman/President of the CMTA, presented a recognition award to Dr. Peter DeJonghe. Dr. DeJonghe traveled from Belgium to present the plenary address at the North American CMT Consortium and helped the group get organized for future meetings. See front page for the full story. Next year, the meeting will be held in Belgium, hosted by Dr. DeJonghe and his wife. Each year after, the meeting will alternate between Europe and North America so that the momentum begun in Canada will continue.

Support Group News

■ California—North Coast Counties

The group met on March 8, 2003 at leader Freda Brown's home. The speaker could not attend, so they had a discussion of "living with CMT." Attendance has been running from five to ten people per meeting. The question arose whether to continue the group or close it down. More people are needed to help with setting up programs and getting out meeting notices. Learning about CMT from others who are having similar experiences is one of the most important values of the group. Let Freda know your feelings about the group, as this may have been the last official meeting. You can call her at 707-573-0181.

■ Kentucky/Southern Indiana/Southern Ohio

The March 22nd meeting of the group was well attended with seven members and four spouses. The new leader, Martha Hall, presented a program in which the group discussed where they might want to go in the year ahead. Meeting locations, frequency of meetings, and topics for speakers were among the things decided upon. The discussions were spirited and meaningful and should help the group to grow.

■ New York—Westchester County/Connecticut (Fairfield)

The March meeting of the support group honored Yvonne Bloch Lugo for her twelve years of dedication and hard work for the Westchester group. In her years of service, Yvonne has handled all correspondence for the group, including the monthly meeting notices and membership dues.

Yvonne has been a great source of inspiration to all of the members, always keeping a smile on her face even during some difficult challenges in her own life.

The group thanks Yvonne for a job well done.



Yvonne Bloch Lugo was the guest at a "thank you" party at the last NY support group meeting.

■ New York—Horseheads

The group held its first meeting at the new location, the Horseheads Free Library on February 12, 2003. There were fourteen people in attendance. The speaker was Sharon Saxe from the Social Security Administration. She discussed filing procedures for Social Security Disability as well as SSI benefits. The discussion was very timely as a few of the participants were just beginning the process.

■ Minnesota—Benson

The spring meeting of the support group was an exciting one, with Dr. Gareth Parry in attendance to give information on CMT and to answer questions. There were 13 patients with CMT in attendance and 8 support people. Dr. Parry discussed the need to get an early diagnosis so that the patient can get braces (which doesn't seem important when you are young, but which will make a big difference to your body as you age) and because it should be known and revealed before marriage. He has seen many divorces occur because a child is born affected by CMT and the unaffected spouse doesn't understand and can't work through the situation.

There is interest in starting a support group in the metro (Minneapolis/St. Paul) area. If that group gets going, the Benson group will probably disband because so many more people can be reached from the city.

NEW SUPPORT GROUP FORMS!

The Northwestern Pennsylvania CMTA support group held its first meeting on Saturday, April 12, 2003 at the Blasco Memorial Library, 160 East Front Street, Erie, PA. The group shared information, concerns, and advice about living with CMT and will feature medical professionals who can answer questions about the nature of the disease.

For the first meeting support group leader, Joyce Steinkamp, BS, CCP, interacted with attendees to build a network of information and support. Joyce is a 15 year veteran of the health care industry with a degree in biology. She has worked in the ICU and operating room environments. Joyce learned of her diagnosis of CMT 22 years ago at age 14 and has taken an active role in dealing with her CMT. If you did not receive an invitation to the group, you can contact Joyce at 814-833-8495 or by email at joyceanns@adelphia.net. ★

CMTA Support Groups

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

Arkansas—Northwest Area

Place: Varies, Call for locations
Meeting: Quarterly
Contact: Libby Bond, 501-795-2240
E-mail: charnicoma57@yahoo.com

California—Berkeley Area

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

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

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

Colorado—Denver Area

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

Kentucky/Southern Indiana/ Southern Ohio

Place: Lexington Public Library,
Northside Branch
Meeting: Quarterly
Contact: Martha Hall,
502-695-3338
E-mail: marteye@mis.net

Massachusetts—Boston Area

Place: Lahey-Hitchcock Clinic,
Burlington, MA
Meeting: Call for schedule
Contact: David Prince, 978-667-9008
E-mail: baseball@ma.ultranet.com

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: Baptist Healthplex,
102 Clinton Parkway, Clinton,
MS
Meeting: Quarterly
Contact: Flora Jones, 601-825-2258
E-mail: flojo4@aol.com

Missouri/Eastern Kansas

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

Missouri—St. Louis Area

Place: Saint Louis University
Hospital
Meeting: Quarterly
Contact: Carole Haislip,
314-644-1664
E-mail: c.haislip@att.net

New York—Greater New York

Place: NYU Medical Center/
Rusk Institute, 400 E. 34th St.
Meeting: 2nd Thursday of each month
Contact: Dr. David Younger,
212-535-4314,
Fax 212-535-6392
Website: www.cmtnyc.org

New York—Horseheads

Place: Horseheads Free Library on
Main Street, Horseheads, NY
Meeting: Quarterly
Contact: Angela Piersimoni,
607-562-8823

New York (Westchester County)/ Connecticut (Fairfield)

Place: Blythedale Hospital
Meeting: 3rd Saturday of each
month, excluding
July & August
Contacts: Diane Kosik, 914-937-2013,
Beverly Wurzel,
845-783-2815
E-mail: DianeK319@optonline.net or
cranomat@frontiernet.net

North Carolina—Archdale/Triad

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

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

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

Ohio—Greenville

Place: Church of the Brethren
Meeting: Fourth Thursday,
April–October
Contact: Dot Cain,
937-548-3963
E-mail: Greenville-Ohio-CMT@
woh.rr.com

Oregon/Pacific NW

Place: Portland, Legacy Good Sam
Hospital, odd months
Brooks, Assembly of God
Church, even months
Meeting: 3rd Saturday of the month
(except June and Dec.)
Contact: Jeanie Porter,
503-591-9412
Darlene Weston,
503-245-8444
E-mail: jeanie4211@attbi.com or
blzerbabe@aol.com

Pennsylvania—Northwestern Area

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

Pennsylvania—Philadelphia Area

Place: University of PA, Founders
Building, Plaza Room A
Meeting: Bimonthly
Contact: Amanda Young,
215-222-6513
E-mail: stary1@bellatlantic.net

Pennsylvania—Johnstown Area

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

The CMTA Report

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

MEDICAL ALERT:

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

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

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

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



What is CMT?

- ... is the most common inherited neuropathy, affecting approximately 150,000 Americans.
- ... may become worse if certain neurotoxic drugs are taken.
- ... can vary greatly in severity, even within the same family.
- ... can, in rare instances, cause severe disability.
- ... is also known as peroneal muscular atrophy and hereditary motor sensory neuropathy.
- ... is slowly progressive, causing deterioration of peripheral nerves that control sensory information and muscle function of the foot/lower leg and hand/forearm.
- ... causes degeneration of peroneal muscles (located on the front of the leg below the knee).
- ... causes foot-drop walking gait, foot bone abnormalities, high arches and hammer toes, problems with balance, problems with hand function, occasional lower leg and forearm muscle cramping, loss of some normal reflexes, and scoliosis (curvature of the spine).
- ... does not affect life expectancy.
- ... has no effective treatment, although physical therapy, occupational therapy and moderate physical activity are beneficial.
- ... is sometimes surgically treated.
- ... is usually inherited in an autosomal dominant pattern, which means if one parent has CMT, there is a 50% chance of passing it on to each child.
- ... Types 1A, 1B, 1D (EGR2), 1X, HNPP, 2E, 4E, and 4F can now be diagnosed by a blood test.
- ... is the focus of significant genetic research, bringing us closer to answering the CMT enigma.

The CMTA Report

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



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