Charcot-Marie-Tooth Association

THE CMTA REPORT

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

CMT Hand Surgery Options

by Barry P. Simmons, MD
Brigham Orthopedic Associates, Inc.

The hand is a complex organ, whose function is dependent on an intricate interrelationship of bones, joints, ligaments, capsules, tendons and nerves. The fingertips have the richest nerve supply of any area in the body. The ability to move our joints in a synchronous and fluid manner requires that all of the structures be intact.

From an anatomic point of view, it is easiest to think of the hand as having two separate muscle units. The small muscles within the hand itself, called the intrinsic muscles, are responsible for balancing the pull of the larger muscles, called the extrinsic muscles, which are found in the forearm. Charcot-Marie-Tooth Disease affects the function of the nerves themselves. Since the function of the muscles depends on the nerve supply to the muscles, then obviously muscle function is also altered. The hand is typically involved in one-half to two-thirds of patients with Charcot-Marie-Tooth Disease and this involvement usually starts after involvement of the lower extremities. This most significant abnormality is usually in the small muscles of the hand, although the extrinsic muscles of the forearm can be involved later. Observing the hands of patients with Charcot-Marie-Tooth Disease, one notices the loss of muscle bulk in the muscles of the hand as well as in the thumb. As well, there can be loss of sensation due to the nerve disease.

Barry P. Simmons

The average age of onset is usually at about 13 years of age with a range of 1 to 59 years. The major complaint is loss of strength or loss of dexterity. Loss of sensation occurs in a much smaller percentage of patients.

Deterioration and loss of strength is slowly progressive, but occurs most significantly in the first 5 to 10 years after the onset of the hand involvement and then remains relatively stable. The most significant involvement, as noted, is in the small muscles of the hand.

As for the hand deformities, classically one sees clawing of the fingers meaning that the joint at the base of the fingers (metacarpophalangeal joints) are overly extended and the joints in the middle of the fingers (proximal interphalangeal joints) are flexed. (Figure 1,2.) Also, the thumb, instead of lying in opposition to the fingers tends to lie flat in the plane of the hand. This deformity results because of the mechanical imbalance of the small muscles of the hand and the large muscles of the forearm.

Surgery is designed to try to correct this imbalance. Since one's usual muscles are not functioning normally, then the goal is to take a muscle that functions in another area and transfer it so as to have it correct the deformity. This procedure, called a tendon transfer, is obviously not

(cont'd on page 2)

Snow, Ice, Floods, and Other Disasters of the Winter

Yes, here we are once more apologizing for the lateness of this issue of the newsletter. The computer is working fine, but getting into the office has been a trick this winter. Not only has the snow and ice kept everyone in the Northeast at home, but when the great thaw came in January, the building in which the office is located was flooded by an overflowing creek. When it rains, it pours... or something along those lines. So, please forgive us once more for the delay in having your newsletter to you.

IN THIS ISSUE:

The Hand- cont'd from pg. 1

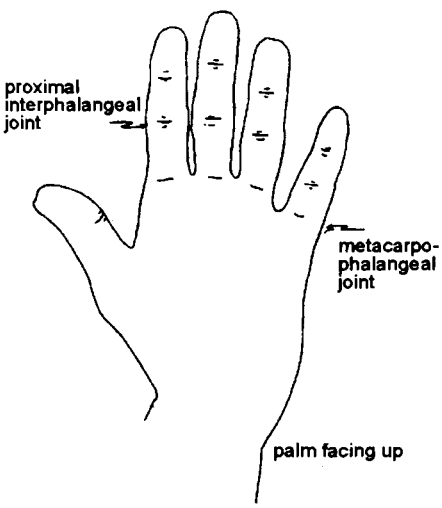

Figure 1

without a price. First, there is always some loss of function, usually strength, when a muscle is transferred from one area to another. Second, as well as tendon transfers can be made to work, they are never quite as good as the original muscles/tendons. Third, the surgeon is faced with the problem of how tight or loose to make the transfer as the subsequent interplay will depend on this amount of tension. Sometimes the transfer has to be "tightened" or "loosened." Furthermore, since this is a progressive disorder, one cannot always be sure that the muscle that is transferred, even if it functions well when the surgery is initially done, will continue to function normally. It is conceivable that, with time, that muscle will also be affected by Charcot-Marie-Tooth Disease.

Thumb function is somewhat less complex. The goal is to transfer a muscle to allow the thumb to be brought around in opposition, i.e. to have the thumb tip face the tip of the index and middle fingers. The problem with that transfer is, as noted in the problem with the digits, whether the muscle that is transferred will be strong enough, tight enough, and continue to function.

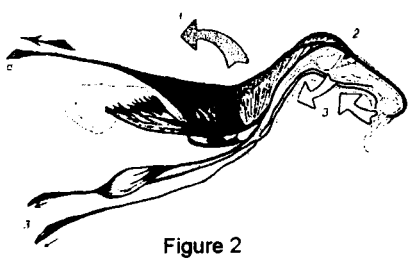

Figure 2

Tendon transfers, as one might imagine, are complex. An example is taking a tendon to flex the metacarpophalangeal joints and extend the proximal interphalangeal joints. This might be accomplished by releasing one of the two tendons that flex the fingers from their attachment in the finger and reattaching them at the base of the finger.(Figure 3) An alternative procedure is to use a tendon that usually extends the wrist, making it longer by sewing an extra tendon from the leg to it and then reattaching it at the base of the finger (Figure 4). The goal of both of these is to have this transfer flex the metacarpophalangeal joint and allow other muscles to extend the proximal interphalangeal joints. For the thumb, one can use a tendon that flexes the ring finger and reattach it to the thumb.

Figure 3

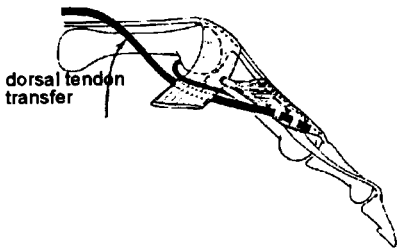

Figure 4

Unfortunately, some patients have deterioration of nerve function sufficient to alter their sensation. There are no surgical procedures that can be done to correct that disorder.

What can a patient expect from tendon transfers? Hopefully, it will allow sufficient re-balancing of the muscles in the hand, so that one's dexterity will be improved and, with better positioning of the fingers, strength as well. However, it is unreasonable to expect there will be a marked increase in strength as a result of the transfers. The major goal is to correct the deformities and allow increased dexterity.

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Baylor Announces CMT Clinic

Editor's note: *The following letter was received in the office of the CMTA addressed to Karol Hitt, CMTA president, just as we were about to go to press. We are very pleased to announce that a major American medical center has established a clinic for CMT patients. This is one step toward the ideal which would have CMT centers around the country so that every patient would have access to CMT medical experts in his/her region. The Baylor Center will be a wonderful resource for people in the South Central part of the country.*

Dear Karol;

I am pleased to inform you that we are beginning a new CMT clinic in the department of Neurology at Baylor College of Medicine. Dr. Richard Armstrong and myself will be running the clinic which will meet on alternate months in the Neurosensory Building at Methodist Hospital. Dr. James Lupski will assist us from the genetic aspect and we are also enlisting the aid of a foot surgeon and an orthotic expert for special bracing problems.

Our plan is to see approximately four to six new families at each clinic. Our first clinic will be on Friday, February 11, 1994 and will run from 1 - 5PM. Facilities for nerve conduction screening of patients and relatives will be available as well as any blood that needs to be drawn for genetic screening.

If the patient load requires more frequent clinics, we can have them on a monthly basis, but at present we feel that every other month will be adequate. For your records, the referral number for clinic appointments is 713/798-5983 and they can ask to be scheduled for the Baylor CMT Clinic.

Sincerely yours,

James M. Killian, MD
Professor and Vice Chairman
Baylor College of Medicine

Items of Interest

Item 1: CMTA Functional Disability Survey Recipients: If you still haven't returned your survey, we ask that you complete it and send it in as soon as possible. EVEN IF YOU ARE MILDLY AFFECTED and you feel many or all of the questions are not relevant to you, we strongly urge you to complete the survey. We want the entire CMT population represented in the results.

Please add the following question to the bottom of the survey:

Please check any categories for which you usually need HELP FROM ANOTHER PERSON:

___ hygiene ___reach ___gripping and opening things ___ errands and chores.

You may mark which categories you need help in or if none apply, write "none."

Through your cooperation we can begin to learn more about CMT and how it affects your life. For those of you who have already returned the survey, a sincere thank you. —*Diana Eline*

Item 2: A book worth considering for the pregnant woman is Mother-to-Be: A Guide to Pregnancy and Birth for Women with Disabilities by Judi Rogers and Molleen Matsumura. 1991. Demos Publications.

$ 24.95 in paperback and $ 39.95 in hard cover.

Item 3: Another book which just came into the office and is unbelievably complete is called 1994 National Edition of the Healthcare Resource Directory. It is an information guide to American medical and social services. The book lists national offices of disease related organizations, professional organizations and self-help groups. In addition, there are U.S. Department of Health and Human Services numbers, State Department of Health numbers and definitions of diseases and disorders along with an alphabetical list of organizations. The directory is the most comprehensive resource directory we've seen to date. It is published by Medical Productions, 713-666-8637. If you order the book, please tell the company where you found out about the book. They will donate $ 5 per book to the CMTA and send the book directly to you.

Item 4: Please remember to inquire if your company has a corporate matching gift program. Depending on the requirements, it is possible that your gift to the CMTA will be matched in full. We are a 501(c)(3) federally approved charity and qualify for most matching funds.

Item 5: Remember that the blood test for diagnosing CMT Type 1A found on chromosome 17 is available from Genica Pharmaceuticals. They can be reached by calling 1-800-394-4493, ext. 106. Ask for Sarah Quiry, customer service representative. A physician must order the shipping kit. The cost of the test is $395.00. §

The CMTA offers its congratulations to Mr. and Mrs. Hart Wurzburg who recently celebrated their 50th wedding anniversary. The following persons joined us in recognizing this wonderful occasion with gifts in honor of the Wurzburgs.

Mr. and Mrs. Robert Logan
Peggy and Lew Pollock
Mrs. Arthur Adler, Jr.
Mr. and Mrs. Sam Lawton
Mr. and Mrs. Richard Wile
Mrs. Edward Goodkind
Mrs. Hugo Sonnenscheim
Ann and Fred Ullman
Roslyn Orlin
Mr. and Mrs. Joseph Lelewer
Mrs. Henry Kahn
Betty Frank
Mr.and Mrs. Edwin Kirchheimer
Frances and Julius
Charlotte Lewis
Shirlie and Mike Goodfriend
Mr.and Mrs. Richard Simon
Mary Jean Field
Mr. and Mrs. Leonard Pfaelzer
Mr. and Mrs. Arnold Wolff
Mary & Floyd Laber
Mrs. Wilbur Grodin
Mrs. Mariette Kaufmann
Mr. & Mrs. Leonard Pfaelzer
Mr. & Mrs. Milton Minkin
Mike Wurtzburg
Mrs. Sidney Robinson
Jeanne & Victor Gidwitz

Alert - Alert - Alert

As of January 8, 1994 our area code changed. Our new telephone number: (610) 499 -7486

Recent CMT Scientific Publications

"Gene Dosage as a Mechanism for a Common Autosomal Dominant Peripheral Neuropathy: Charcot-Marie-Tooth Disease Type 1A", Roa, Garcia, Wise, Anderson, Greenberg, Patel and Lupski. The Phenotypic Mapping of Down Syndrome and Other Aneuploid Conditions, pages 187-205. 1993 Wiley-Liss, Inc.

"Molecular Basis of Charcot-Marie-Tooth Disease Type 1A:Gene Dosage as a Novel Mechanism for a Common Autosomal Dominant Condition." Roa, and Lupski. The American Journal of the Medical Sciences, September 1993. Vol.306, No.3.

"Inherited Primary Peripheral Neuropathies:Molecular Genetics and Clinical Implications of CMT1A and HNPP. Lupski, Chance, and Garcia. Journal of the American Medical Association, November 17,1993, Vol.270, No.19.

"Connexin Mutations in X-Linked Charcot-Marie-Tooth Disease", Bergoffen, Scherer, Wang, Scott, Bone, Paul, Chen, Lensch, Chance, and Fischbeck. Science, Vol.262, December 24,1993.

"Nerve conduction studies in Charcot-Marie-Tooth polyneuropathy associated with a segmental duplication of chromosome 17" Kaku, Parry, Malamut, Lupski, and Garcia. Neurology, September 1993.

"Dejerine-Sottas syndrome associated with point mutation in the peripheral myelin protein 22 (*PMP22*) gene" Roa, Dyck, Marks, Chance and Lupski. Nature Genetics, vol.5, November 1993.

"Evidence for a recessive *PMP22* point mutation in Charcot-Marie-Tooth disease type 1A" Roa, Garcia, Pentao, Killian, Trask, Suter, Snipes, Ortiz-Lopez, Shooter, Patel, and Lupski. Nature Genetics, Vol.5, October 1993.

"Molecular Analyses of Unrelated Charcot-Marie-Tooth (CMT) Disease Patients Suggest a High Frequency of the CMT1A Duplication" Wise, Garcia, Davis, Heju, Pentao, Patel and Lupski. American Journal of Human Genetics, 53:853-863, 1993.

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Researchers Lupski and Chance Study A Baffling Genetic Disease - Their Own

by Dennis Breo

(This article is reprinted by permission of the American Medical Association. It appeared in The Journal of the American Medical Association, November 17,1993, volume 270, NO.19, pgs.2374-2375.)

Charcot-Marie-Tooth disease (CMT), a progressive neuromuscular disorder of variable severity, is named after the French and British neurologists who first described its symptoms of muscle weakness and wasting in the feet and hands in 1886. Today, CMT affects one of every 2500 Americans, or 150,000 people, and is the most common inherited peripheral neuropathy.

The search for its causes has led to cutting-edge genetic findings, notably by two rising young researcher stars - James R. Lupski, MD, 36, a pediatrician and medical geneticist at the Institute for Molecular Genetics at the Baylor College of Medicine in Houston, Texas, and Philip F. Chance, MD, 40, a pediatric neurologist at The Children's Hospital of Philadelphia and the University of Pennslyvania.

Their latest findings are described in an article published in this issue of JAMA, but what is not revealed is that their personal interest in the disease is intensely personal - both men have suffered, both physically and psychologically, since their early teens from this often-misunderstood disorder.

Chance gave up a potential career as a clarinetist because of weakness of his hands and now walks with the aid of plastic braces; beginning at age 14, Lupski endured 11 surgical procedures on his feet to enable him to walk with difficulty, and he now suffers from arthritis. Colleagues, competitors -and friends- Lupski and Chance both agree that Charcot-Marie-Tooth is a terrible name for their affliction. The two researchers took time out from their activities at last month's meeting of the American Society of Human Genetics (ASHG) to discuss their unique perspective on CMT with this reporter.

Chance, a quiet, reserved, slender man who is publicly discussing his ailment for the first time and who is given to wry understatement, notes. "I do tell my patients that Charcot-Marie-Tooth disease has nothing to do with the teeth!" Lupski,

Philip F. Chance

a big, bearded man who is given to effusive outbursts of enthusiasm, adds, "The details of this are not for publication, but I once had a grant application returned with the notation, 'Your request for research on tooth decay has been approved!'"

Both researchers have certainly done their part to explain the disease. In 1991, working with blood samples drawn from a cluster of large Cajun families in southern Louisiana who were being treated for CMT, Lupski and his Baylor team indentified an area on chromosome 17 in which a portion of DNA was duplicated, leading to a triple dose of genetic material, rather than the normal two. This erroneous triple helping of DNA - "too much of a good thing," in Lupski's words - causes 85% of all cases of CMT.

Lupski calls the discovery "a completely new mechanism for human disease." The finding opened up the possibility that DNA duplication, rather than gene mutations, might hold the answer to such devastating genetic disorders as Alzheimer disease, schizophrenia, and even breast cancer. Instead of looking for a needle in a haystack - the chance mutation of one of the 50,000 to 100,000 human genes - researchers were alerted to the possibility of simply finding too much hay. Chance calls this 1991 discovery by Lupski "totally novel" and the "premier observation in genetics during 1991."

Nevertheless, Lupski had to scramble to even get his results published (in *Cell*,1991;66:219-232), and they were not found worthy of making the ASHG meeting agenda that fall. "To receive proper credit," Chance says, "observations about a little-known disease like CMT have to be 10 times as important as most other genetic findings." Lupski says, "CMT does not get the respect it deserves."

There are 30 to 50 genes in the duplicated portion of DNA on chromosome 17 that causes CMT, so Lupski and his team stepped up their efforts to isolate the culprit gene. This summer, he reported (in the *New England Journal of Medicine*, 1993;329:96-101) that a special gene in the duplicated material - *PMP22*, or the peripheral myelin protein of molecular weight 22 - causes CMT in mice when the gene is mutated. He now says, "The *PMP22* gene appears to be the major player in causing CMT."

In the meantime, Chance and his research colleagues have identified a gene on chromosome 1, changes in which also lead to a form of CMT. And Chance discovered that deletion of DNA material on chromosome 17, a reciprocal phenomenon to the duplication that causes CMT, is known as hereditary neuropathy with liability to pressure palsies (HNPP). The genetic causes of these inverse diseases are reported in this issue of *JAMA*, an event that moves Lupski to observe, "I'll be honest with you - this is a great paper!"

Instead of looking for a needle in a haystack... researchers were alerted to the possibility of simply finding too much hay.

Lupski sums up, "We've learned more about CMT in the last 2 years than in the previous 100. Still, I would say that we're only at 5 on a scale of 10 in terms of understanding this disorder. We won't be satisfied until we're able to offer therapy to patients." Chance adds, "We've learned a lot but I can't sit down with my patients and write a prescription to help them."

Interestingly, the two researchers have yet to solve their own cases of CMT. The DNA duplication and point mutations in *PMP22* account for 90% of all CMT cases, with the remaining 10% caused by changes on four other chromosomes: 1,

(continued on the next page)

8, the X chromosome, and an unknown fourth chromosome.

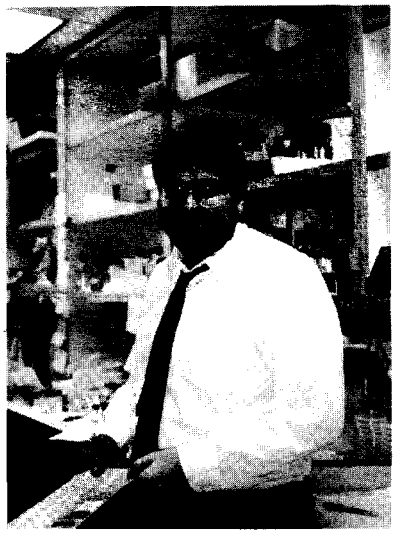
"The very day we found the DNA duplication on chromosome 17," Lupski says, "I had my blood drawn, but there was no duplication. When we discovered the point mutations in the *PMP22* gene, I had my blood checked, but there was no mutation. And when Phil's lab discovered the CMT-causing gene on chromosome 1, I also had my blood checked, with no luck. Neither of my parents had CMT, but four of their eight children do, so my inheritance is obviously due to a recessive gene. I may never find out what caused my disease."

Chance says, "Jim and I have a bet for dinner on who will find his cause of disease first, and I think that Jim will have to pay. I'm getting very close to proving that my case is linked to genetic errors on the X chromosome."

Whatever the cause, CMT made the young Chance and the young Lupski suffer, and there were few places to turn for help. As he draws a diagram on a blackboard to explain to this reporter the reciprocal DNA duplicaton/deletion that is the common cause of CMT and HNPP, Chance drops the chalk. Several times. He sighs, as he recalls:

"My case was very poorly handled. My parents and friends (in Memphis, TN) were not terribly sophisticated medically and there was a tendency to ignore my condition. With CMT, you have trouble picking up your feet and walking normally. It's often called a 'slap-step.' You get tired very quickly and you can't play sports at all. Later, your hands become weak and you have trouble with zippers and doorknobs. It's very embarrassing and distressing, especially when you're a teenager.

"To keep from tripping over your toes, you tend to raise your feet high into the air, almost as if you're marching. Mother used to yell at me,'Pick up your feet and walk normally!' as if I were walking funny just to annoy her. Even today, mother and I don't talk about my condition because of those early days. I ignored my problem until my high school physics teacher intervened. She was the daughter of an orthopedic surgeon and insisted that I pay him a visit. There ensued a variety of unnecessary, expensive, and painful tests - myelograms and spinal taps - to rule out damage to my spinal cord. Finally, I was seen by a neurologist who performed nerve conduction velocity tests to make the diagnosis. Then, all the doctors I had seen just



James R. Lupski

dropped my case. Apparently, there was nothing more to be done."

Chance literally tiptoed through medical school (at the University of Tennessee, Memphis),"shambling about with great effort and great fatigue. My classmates and teachers thought I probably had had polio when I was younger, but nobody ever said anything. After all, this was the South and people simply don't ask. Plus, I was into denial, not wanting to admit that I was impaired.

"This pattern of denial persisted until I began a fellowship in medical genetics at the University of Utah School of Medicine. My mentor, Fred Ziter, MD, insisted that I do something and I was soon fitted for braces. My, was that a rebirth. Suddenly, I could walk almost normally. It was one of the greatest things ever."

By then, however, Chance had turned his back on his first love- music. As the first clarinetist in the Memphis Symphony, he had once planned a career as a professional musician, but it was not to be. "I was a very good clarinetist," he says, "and I have the tapes to prove it, but I knew that the advancing weakness in my hands wouldn't allow it and that medicine would be a better choice for me in the long run."

Looking back, he says, "Of course, I should have been referred to a muscular dystrophy clinic as soon as I was diagnosed and been given the options of braces or surgery. It would have saved a lot of pain."

Lupksi's feet were so bad "that I was walking on the sides of my feet and always spraining my ankles. I loved sports, especially football, but there was no way I could ever be any good. I was referred to a Long Island (NY) orthopedic surgeon, who recommended surgery to stabilize the ankles. I spent almost 1 year in a wheelchair after both my feet were operated on, and that gave me a lot of time to think about what I wanted to do with my life. I chose medicine, and in my wildest dreams I never thought that the genetic research I am doing would be as fascinating as it is."

Lupski is quick to credit Carlos A. Garcia, MD, a New Orleans neurologist and the third author of the article on CMT in this issue of *JAMA*, with making possible much of his landmark research. "To do research in this area," he says, "you need the assistance of a clinician who enjoys the trust of his patients. For 20 years, Carlos has run three Muscular Dystrophy Association clinics in southeast Louisiana, and over that time he has come to know a lot of large Cajun families with CMT. He arranged a Cajun barbecue in Houma, LA, and put the word out for all these family members to come and give blood for the 'important research' that my team was doing. That's how we were able to make our 1991 discovery of the DNA duplication."

"Your request for research on tooth decay has been approved..."

Both Lupski and Chance also see CMT patients in their pediatric clinics. "I tell my patients," Chance says, "that this condition will not shorten their life and it will not affect them mentally. And I try to help them make wise career and family-planning choices." Lupski adds, "Right now, we can offer braces and surgery and career counseling and, someday, we may be able to offer genetic therapy to actually regulate the expression of the *PMP22* gene and stop it from doing its damage."

Neither Lupski nor Chance is worried about the risks to their families. Lupski has two very young daughters and says,"I am in favor of presymptomatic testing, and if my wife agrees, will probably have my daughters tested for CMT when the time is right. I don't want them planning a career in ballet if the disease is present." Chance says, "My wife and I want to have children, and the fear of the unknown won't stop us."

Reflecting on all the needless suffering he endured, Chance concludes, "I try to give all my CMT patients one overriding message - hope."

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Patient Profile : Russell Kimball

(Editor's note: People often wonder how we decide whom to profile. Russell wrote us a wonderful note with his contribution and we were so impressed with his words, that we asked him to be the subject of this profile.)

Russell Kimball could impress anyone with his accomplishments and his attitude, but if you factor his having CMT into the picture, it is impossible not to come away in awe of him.

At the age of 33, Russell is the Vice President of the Washington State Jaycee organization, and has been on several youth organization boards of directors. He began a program called the Business Diagnostic Center while working on his MBA because he felt that MBA students needed to have personal interactions with business people in their community. Following the initiation of that program, he was honored with the Faculty Award for Outstanding MBA Student Service. He has begun his own business, Operational Alignment Specialists, which focuses on aligning business operations to an effective core business strategy. He has written a book and developed several business simulation exercises for non-profit youth leadership training programs.

The following are Russell's own words about his life and his attitude.

Russell Kimball

"I did not find out I had CMT until I was almost 17. While growing up I shared the same challenges that most other children with CMT had (I call it the "last picked for basketball" syndrome) where I always seemed to stumble and was very uncoordinated. I will always remember the mean-spirited teasing I received from othe children for being so uncoordinated and being so poor in sports.

However, I, like most other people with the disorder, recognize that CMT is an integral part of who I am. If I did not have this disorder, my personality would be much different. Just prior to my diagnosis, I was able to run a 26 mile marathon in under 4 hours. I trained hard and did not let the fact that I came in dead last in over 90% of the high school track meets I competed in let my confidence waver. Knowing that I have CMT has convinced me to make physical exercise a part of my life. I feel the disease taking its toll on my arms and legs, but refuse to give in to it. I have learned persistence and discipline in all parts of life that most folks don't seem to have. Much of this I attribute to knowing that I need to train twice as hard as everybody else to be half as good.

Coping with CMT has made my personal motto - "you can't lose if you refuse to quit." As an undergraduate, I graduated *Summa Cum Laude* with a degree in Mechanical Engineering. I am 33 now and have had many other personal and professional accomplishments. Do I sometimes get down and feel weak from the disease and other life troubles? Most certainly, but coping with the disease has given me the inner strength to tough it out. Recently, I have completed my MBA and decided to follow my dream, which is to go to work for myself as an independent consultant and trainer. The odds have been against me from the start, and every month I am concerned about simply paying the rent. But somehow, I have survived for 18 months. Progress seems to be agonizingly slow at times, but I sense that if I refuse to quit that I will find a way to overcome."

Russell Kimball is just one more of the impressive numbers of CMT patients who consider life with CMT a challenge and not an obstacle.

The Orthotist Speaks

I have been asked to provide some information about current orthotic appliances, shoes, and orthotic inserts for those people with CMT. Let me first begin by saying that it is important to work with a professional who is familiar with the various levels of care necessary for a client with CMT. In many cases, patients can be helped tremendously by altering their shoewear alone. There are an abundance of shoe manufacturers who sell shoes that can be very helpful. A cross training style of athletic shoe can provide stability for those with ankle weakness. There are more stylish shoes today with deeper toe boxes, and softer leathers for those with early toe problems. A Certified Pedorthist or Orthotist may be able to direct you to some of the proper shoe choices.

Secondly, the field of Pedorthics, which is the designing, manufacturing, fitting and modification of shoes and related foot appliances for foot disorders, is expanding rapidly and can be quite a help to those having foot problems. With the guidance of a physician, a pedorthist can work to stabilize and correct some foot problems that those with CMT are quite familiar with. The pedorthic and orthotic industry are now utilizing revolutionary polymers and silicones to produce orthotic devices that are lightweight, cosmetic, and highly functional. The biomechanics of the foot and ankle is a complex subject and it is important for the practitioner to be well versed in order to properly care for a patient with CMT-related foot disorders. Also, new technologies in shoewear, shoe inserts, and braces are abundant, but it may take some searching for the right person to manage your particular needs. Also, it is important that there be a good understanding between the practitioner and patient.

Good communication combined with newer technologies can only bring the level of care for those with CMT higher and provide the patient with a better chance for comfort and stability.

Roger Marzano, C.P., C.Ped.
Yankee Bionics, Akron, Ohio

Identification of the CMT X Gene

Michael Bennett Phil., D.

In the December 24th issue of Science, Dr. Kenneth Fischbeck (a member of the CMTA Medical Advisory Board) and his colleagues published an article identifying the CMT gene that is found on the X chromosome. The gene's function is to encode for a protein called connexin 32. Connexin 32 forms gap junctions between cells. These junctions contain tiny channels that connect cell interiors and allow diffusion of small molecules and transmission of electrical signals. There are 11 or so related connexins, each encoded by a separate gene, that also form gap junctions. Different connexins are found in different tissues, but one connexin can be found in many tissues and one tissue can contain several connexins.

Connexin 32 is prominent in liver, pancreas, mammary gland, central nervous system myelin and peripheral nervous system myelin. In myelin, gap junctions connect different parts of the same cell rather than different cells. Peripheral myelin is of the most obvious interest to the CMT patient, and there are many questions concerning their symptoms which are amenable to study with normal tissue from human or lower organisms. Why is the effect limited to the extremities when as far as we know myelin is the same close to the spinal cord as it is out in the finger tips? Are there differences in myelin in normal individuals ("wild type" in genetics terminology) in the amount of Cx32 expressed along the axon? These are questions that CMT X patients now make significant. It is quite unknown what Cx32 does for myelin. Finding out might make it possible to ameliorate, reverse, or stop the progression of the disease in CMT X patients.

Scientists are also interested in what Cx32 does in other tissues. Scientists familiar with gap junctions were astonished at the lack of symptoms in most organs suffering loss of function of one of these scientists' favorite molecules. Other connexins normally present in these organs may carry out the duties of the non-functional one (for example, connexin 26 is found in many cells that also have connexin 32). Cells may make other functional connexins in response to lack of functional Cx32. This hypothesis can be tested by looking at affected tissues with techniques that determine the amount of the different connexins present.

Now that the genetic basis of CMT X is known, there is a need to examine other organs and tissues that are known to contain connexin 32 junctions. Clearly, the loss of function does not cause obvious problems. However, subtle changes or reduced ability to respond to adverse physiological circumstances could have been missed by the neurologists to whom the CMT X patient is generally referred.

There is an important potential benefit to the CMT X patient of these investigations. Most directly, he/ she and his/her doctor should know in what respect the patient may be more susceptible than normal. Prior knowledge of how the patient may differ in his/her response to various diseases or drugs should guide treatment of any disease that involves an organ containing connexin 32.

Many CMT X patients are already families studied by "neurogeneticists". If you know your CMT type, it may be important to your treatment.

The Research Fund of the CMTA received $1042.00 in gifts made in memory of Irma Oakleaf. We wish to offer our condolences to her family and to thank publicly those who gave in her memory.

Mr.and Mrs. Todd Moore
Mary Alice Rapp
R.H. and Anne Oakleaf
Clair Schell
Kaiser Aluminum Corporation
Newark Healthcare Center
Mr. and Mrs. Kenneth Mauter
Ross and Marilyn Winegardner
Charles and Martha Prouty
Tim and Pamela BeVier
Louise Dete
Hank and Bernadine Mesewicz
Mary R. Paulsen
Fred Palmer
Dan and Margaret Evans
Debra Sinclai
rMr. and Mrs. S.V. Jackson

Wanted.... CMT X Patients

Dr. Michael V.L. Bennett, Chairman of the Department of Neuroscience at the Albert Einstein College of Medicine, has asked the CMTA to do a call for participants. Dr. Bennett and his research group need small samples of various tissues from diagnosed CMT X patients; that is, patients whose CMT is caused by the CMT gene located on the X chromosome.

At this early stage of their studies, they need small tissue samples from CMT X patients to compare with normal tissue. If you are a CMT X patient who is undergoing a planned operative procedure in which tissue is to be removed and you are willing to donate a fraction of that tissue, please contact Dr. Bennett or have your physician contact him. Proper preservation of tissue is critical for subsequent study. Relevant procedures include liver or breast biopsy, gall bladder removal, mastectomy, brain surgery for epilepsy and removal of Schwannonmas. (Autopsy material with a short postmortem interval would also be invaluable.) Examination of these tissues promises NO immediate therapeutic benefit. It may reveal how CMT X patients do or don't compensate for loss of Cx32 and ultimately lead to new therapies.

Dr.Bennett's group is also planning neurological workups measuring peripheral nerve conduction proximally and distally, central nerve conduction, and visual function. Initial studies will be done in the Bronx. Transportation will be provided for patients in the greater metropolitan area for measurements involving a few minutes to a few hours of their time.

Dr. Bennett's address is: Department of Neuroscience, Albert Einstein College of Medicine, New York, NY 10461;telephone: 718/430-2536, FAX: 718/824-3058.

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.

Memorials

In Memory of	By
William Weniger	Mae DiMaio
Edith McCormick	Margaret Wegehoft
Joan-A beautiful human being	Reeva and Scott Sterling
W.J.Wheat, Jr	Marsha Wheat
Nancy Westerfer Smith	Catherine Ebersole
Ruth Nellis Potter	Mary Elizabeth York
Ralph Binford	Marilyn Dodge
Marguerite Henry Criner	Frieda Schroder
Dr. Chase Jones	J.F. Boyce
	Mary and John Pickett
	Mark and Nancy Hillis
	Edward Davidson
	The Kingsbury Temperance Fund
	Kent and Peggy Schoneman
	Dale and Dorothy Meecham
Tom Calabro, Sr	American G.I.Forum, Omaha Chapter
	Albert Vaillancourt
	Dudley and Kay Allen
	Gladys LaMaster
	Anton & Katherine Miratsky,Jr
	Richard and Carolle Peklo
	Joseph and Pamela Inserra
	Mr and Mrs Frank Ferro
	Joe and Nancy Conley
	Willard Urban
	Al and Connie Naticchioni
	John and Mary Sporcic
	Cliff Shepoka and family
	Allan and Mitzi Bogacz
	Rose Povondra
	Mark and Coleen Driscoll
	Louise Driscoll
	John and Darlene DiBiase
	Bob Lenahan
	Bud & Josephine Kruegers
Murray Smith	Lurlye Smith Comegys
Martin Edelheit	Ruth Edelheit
Sophie Jacobson Adler	Ruth Edelheit
Dorothy Fox	Sylvia Kauffman
	Barbara Bichel
Sabina Dunstan	Dorothy Dunstan
Yvonne Simnick	Ashel Miller
Irene Weiss	Charlotte and Paul Brieff
Jessie Johnson	Minna Sand
Jessie Cunningham	Harold and Dorothy Cunningham
Jessie Cunningham	Dr. and Mrs. L.B. Cunningham
Jessie Cunningham	Mike and Carol Joyce
Jack Goldman	Doris Goldman
Roland Smith	Margaret Smith
Robert Tidball	Elinor Tidball
Mother	Reeva Sterling
Harry Schaller	Victor Schaller
Isabel Sprague	The Moores
David Friedman	Herbert Friedman
Mildred Winchell	Phyllis Warren
Josephine O'Grady	John O'Grady
Kathleen Chute Hausman	Martha Bowman
Lotti Soellner	Trudi Kayser
Anne Boca Kerrigan	Michael Schwartz
Brendan Kerrigan	Michael Schwartz
Mollye Schwartz	Michael Schwartz
Rena Brunetti	Richard Brunetti,Sr.
James Vorhees	Vicki Vorhees
James Vorhees	Betty Ann White
James Vorhees	Evelyn Bogues
Harry F. Schaller	Harry P. Schaller
Shafiqua Atalla	Yacoub Atalla
James Vorhes	Mr. and Mrs. John Marakas
James Vorhes	James Vorhes, Jr.
Hy Goldfarb	Morris and Loretta Woolman
Hy Goldfarb	Mr. and Mrs. Isadore Miller
James O'Neil	Grace Wangaard
Michael McClellan	Gregory and Lori Ann Schrad
Emily's Grandmother	Susan Louer
James Vorhes	Chris Gram and Bob Fontichiaro
Beloved Mother of Larry Stoller	Shirlee Bank
Beloved Sister	Shirlee Bank
James Vorhes	Doris Manvel
Rivian Stoller	Jerry and Debra Bank
Bill Claussen	Brian and Nancy Hogan
Elizabeth Berg	Irving and Bethel Bank
Margaret Dailey	Karol Hitt

Honoraria

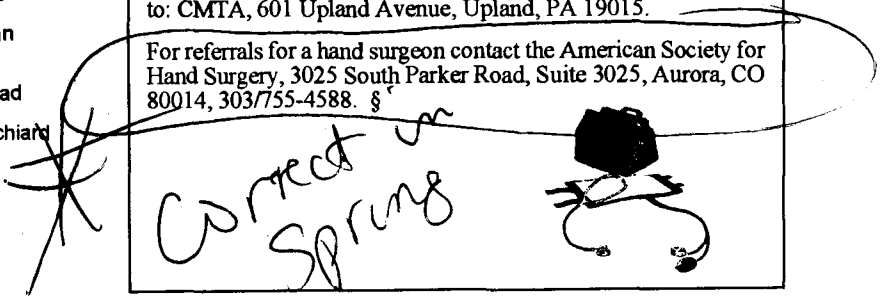
In Honor of	By
Lee Sullivan	Mary Beth Tamulevich
Hugh & Sue Willis	Susie Willis
Brett Nowel	Clyde Goodrich
Iris Golmetz	Karen Gabel
Debra Czarnecki	Carol Fink
Rebecca Sand	Rhoda Sand
Jason Everhart	Peggy Jumper
Charles & Tom Lynch	Christopher Lynch
"Cliffy"	Karen Weiland
Alexandra Segal	Dori Morales
	Richard & Louise Goodman
	Diane Berman
	Len Ferretti
	Linda Reff
	Art Taxman
	Louis & Hayley Higgins
Mrs. Elizabeth Petty	Mr. and Mrs. Douglas Moody
Mr. and Mrs. Henry Eldrege	Mr. and Mrs. Douglas Moody
Robert Brieff	Charlotte and Paul Brieff
Jane Terranova	Charlotte and Paul Brieff
George Phy	Elizabeth Oliver
Diane Higgins	Joseph Higgins
Alex Segal	David and Geraldine Freedman
Philip Magnus	Susan Louer
Christopher Lynch	Charles and Helen Lynch
Brett Nowel	Clyde and Ruby Goodrich
Jean, Jennifer and Joy Rios	Joe Ellenbogen
Stephen Schaefer	Margaret Schaefer
Buzz Van Almen	Arla Van Almen
George Smith's birthday	Leonore Smith
Teri Daino	Patricia Pesci
Tom Lynch	Charles Lynch
Diana Davidson	Jean Waldron
Rebecca Sand	Sybil Whitman
Rebecca Sand	Judith Goldman
Charles T. Lynch	Charles and Helen Lynch
Ruth Wendkos	Mrs. Fred Bandler
Aaron Greif	Nina Bernstein
Judy and John Grocott	Robert Hawkes
Mr.and Mrs. Stuart Feen	Ruth Feen
Elsa Johnson	Tom and Kathy Kroger
Irving Schwartz	Michael Schwartz
Elia Atalla	Yacoub Atalla
My grandchildren	Alice Marie Stout
Henry Friedmann	Peter Friedmann
Mrs.Peggy Jumper	Gary and Dianne Everhart
Christy Wagner	Pamela Gatfield
Geoffrey Clarkson	Elizabeth Clarkson
Gary Rubenstein	Margo Itule

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. §



Letters to the Editor

Dear CMTA:

I appreciate the correspondence that I have received lately from the CMT Association. I was diagnosed 15 years ago with CMT and this is the first that I have heard of your association and that was strictly by accident. You hit the nail on the head in your letter with the "lack of knowledge regarding CMT on the part of medical professionals." I had been left for "cripple" by the doctors but took it upon myself to defy this disease. I would certainly like to help in any way that I can but in turn, I was wondering if you could help me by taking a moment to read my story.

I am 34 years old. I have always attended a gym because the doctors told me simply to keep exercising and not become stagnant. One and one-half years ago I joined Gold's Gym and decided to work with a personal trainer. I told my trainer my physical disabilities and his attitude was "if you can walk, you've got muscle. You got muscle,we can build it." Slowly, but surely I became increasingly stronger and some people even felt my gait was a little better. My self esteem was so increased by this. I wanted to continue forever and call Jerry Lewis of MDA and tell him my progress. I then contacted MDA,locally as well as nationally, told them of my progress in hopes of getting some kind of funding towards this "one-on-one" training. Basically, their answer was "that's nice that you are doing so well, but unless you are crippled, we can't help. We do not aid such things." I knew no where else to turn.

Five months ago, I went for a routine check-up to my neurologist. He basically shot-down my "working out" at the gym. "You could hurt yourself and you need to be careful," he said. Well, I can hurt myself stepping off a curb outside! Through further conversation we discussed testing, to discover whether my CMT was dominant or recessive. This lead me to Shands (hospital). I was told there is no such test, however, 3-5 years from now there may be. This person at Shands, however, lead me to you. He sent me a brochure on CMT which had your VCR lectures listed. I ordered the one on physical therapy. This woman was my miracle. She was the first person to more or less condone what I have been doing. She said, if you have muscle, work it! I finally felt vindicated that everything I had been doing, despite my doctor's advice, was actually good for me. She went into an in depth discussion on muscles and showed different severities of CMT which I was not aware . Obviously, I have been afflicted slightly because I do not have some of the severe problems that CMT can cause shown on the video, or I have arrested the disease enough with my exercising that this has really helped.

My biggest concern to CMT patients is that we do not let our doctors debilitate us mentally with what we cannot do! What about what we can do? I am so much stronger physically and mentally because I went one step beyond what the doctors said. No one ever encouraged me to push myself because by the time I was 20, I would be in braces and/or a wheelchair. Well, at 34, I have neither. Obviously,"the gym" is not for everyone, but I would like to encourage those who still can do strengthening exercises , to try. It is definitely a slow process and needs to be monitored by a professional, but I have found that few physical therapists have dealt with CMT. I am a medical transcriptionist and obviously have access to many physicians. I have met few who are familiar with the disease.

Does the CMT Association have any kind of funding for such things as personalized training in a gym or physical therapy facility? Obviously, you are asking for contributions, so I would assume not and I will send in what I can. Is this something that the Association might want to consider in the future? I agree, with only 5000 members out of 125,000, we definitely need more input. Your letters have already helped me, simply knowing that there is someone out there who understands.

I would like to see those who are diagnosed get physical strengthening help early on in hopes of arresting the disease. I was not diagnosed until age 13 and consider myself very fortunate that it never progressed, considering the fact that my doctors gave me no encouragement and left me in the dark. I have had to educate myself as an adult on CMT. No other family members that we know of have this disease and I am the fourth of five children. I also find it amazing that in 15 years plus, the only literature that is still sent out is on "stretching the muscles." Agreed, this is very helpful, but does not give strength to the muscle.

I don't know how rare or common my story is for I know no one personally who has this disease. I guess I just wanted to vent to someone who possibly understands my situation and spread the word that some of us can be helped to a degree and hope that they are seeking that help.

Please keep up the education and information resources of the CMTA. I commend your volunteer Board of Directors for all of their efforts and time.

L.B. Casselberry, FL

Dear CMTA:

Let me begin by telling you a little about myself. I graduated from Johnson County Community College in 1990 with a degree in Restaurant Management. Currently, I am living and working in the Kansas City area.

In the Spring of 1992, I was diagnosed with Charcot-Marie-Tooth Disease. This diagnosis was a long time in coming because several medical professionals in the Kansas City area were unaware of the disease. Only after moving to Las Vegas was I diagnosed with having CMT.

When I was first told about having CMT, I was quite upset, yet grateful that the problem I had was known. As you yourself know, this can be an incredible shock to a 23 year old man.

I began looking for a therapist who would be willing to work with me on stretching and strengthening exercises. After searching for approximately two months, I found a young woman who brought me new hope and ideas. Because there is not a lot known or at least printed

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Write us: Letters / The CMTA
601 Upland Avenue
Upland, PA 19015
610/499-7486

Letters - continued from p.10

about CMT, many of our exercises were very unique experiments. I immediately began to lift weights five days a week to help strengthen my arms and legs. I decided that this disease would not defeat me and I became very determined to prove it. To my own credit, I never gave up on myself.

Working restaurant management requires standing on your feet for long hours every day. After approximately three to four months of very intense therapy, I began to notice that I could walk faster and more comfortably; also, my forearms began to gain in size and strength. Although I do not require a therapist any more, I will never forget what Michelle did for me. Not only did she teach me physical endurance, but she showed me that there was hope.

On June 21,1993, I had an operation on both feet and ankles. I was in a double cast for six weeks. The operation was well worth it, even though the pain was very intense. It has now been five and a half months since my surgery. I can now use my dorsi flexion on both feet to almost a non negative position, which was not possible before my operation. I realize that full recovery will take awhile, but I am determined that I will defeat this disability.

I continue to lift weights five days a week and do a number of foot exercises three times a week. Although I do not have all of the strength back in my legs after the surgery, walking is now easier than ever before.

I have spoken with other CMT patients and feel very blessed that my condition was not nearly as severe. I realize that each CMT patient has his or her own limitations and the disability may vary from person to person.

The main reason for writing this letter is not to receive sympathy, but to express hope. Every individual is different and each individual has his own limitations, yet with dedication and faith these limitations can be overcome.

I currently work anywhere from forty to fifty-five hours a week. Everyday tasks which were once very frustrating are quickly becoming a standard part of my life. I realize how alone a person can feel when diagnosed with CMT. I am very willing to speak with any CMT patient who would like information on even a little encouragement. If there is any way in which I could be helpful, please contact me at 816-942-1817.

Allan, KC, MO

Dear CMTA,

I'm writing for the sole purpose of perhaps offering a ray of hope and encouragement to others affected with CMT and urge them never to give up in trying to improve their condition. Neurological disorders apparently are very difficult to diagnose, so consult the very best doctors.

As a child, I could compete in some sports but was never good at any of them. In basketball, I could not use thumbs and wrists like other kids. At the time, I was not aware of any disadvantage. Since my parents were divorced, I had not contact with my father, who was the CMT donor. No one called attention to the fact that I had a slight limp. I have two sisters, one with, and one without, CMT.

When I enlisted in the US Navy Seabees in 1942, it was discovered that I could not bend and jump around on my toes. When asked if I had trouble with my feet, I replied that I had not and was accepted as a third class petty officer (electrical).

After serving two years in the southwest Pacific, (then as a first class petty officer) working seven days a week and no leave, I awoke one morning and was so dizzy that I couldn't walk. The doctors discovered that I had no tendon reflexes and asked how I had managed to get into the service! I always knew that I had no knee jerk.

After being examined (no diagnosis) I asked to be returned to my outfit since I thought that we would be returned to the states soon. The doctor complied and shortly we did return to the states. I had a thirty day leave and returned to active duty. I was then assigned to station force duty in California where I was discharged at the end of the war. I received an honorable discharge - not a medical discharge.

I started a small electrical business and in a few years discovered that the muscles between the thumbs and forefingers of both hands were diminishing. Also, both feet were turning inward and I was having difficulty walking.

In 1951, my local doctor sent me to "experts" in the city! I was examined by a neurologist and an internal medicine doctor who arrived at the diagnosis of amyotrophic lateral sclerosis! Of course, my wife and I were devastated. My local doctor suggested that perhaps the experts had not seen a case of ALS and I should go to Barnes in St.Louis or Mayo Clinic in Rochester, MN for confirmation. In 1951, we went to Mayo's and were delighted - after being properly diagnosed with CMT by Dr. Harry Lee Parker.

In 1959, after consulting with Dr. Mulder at Mayo's in Rochester, I had tendon transplants done by Dr. Edward Henderson on both hands to restore thumb and forefinger action. The operation was a great success and I continue to have good use of my hands.

In 1964,my feet continued to deteriorate and I had triple orthodesis performed on both feet. The ankles were then straight, but now I had toe drop. I wore spring wire toe lift braces on both legs for a few years which were cumbersome and often broke.

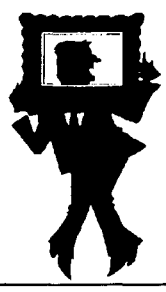
I went to Mayo's in 1973 and talked to Dr. Martin who suggested that I go to "Rochester Orthopedic" to look at a toe lift brace made of a new plastic from Germany. I had a pair of braces made from the new material and wore them 365 days a year for twenty years! A terrific material - no breakage during the entire time.

My knees continued to bend back and I was becoming more unsteady. In 1993, I again went to Mayo's to see if there had been any new developments. I requested to see Mr. Arand Brinks of "Rochester Orthopedics."

Mr. Brinks and his son suggested a leg brace which they had been making for a few years. I decided to take a chance and have a pair made. I am very happy that I did! At first, I was disappointed because of the metal hinge at the ankle. I couldn't use my usual side zipper six inch boots. However, I now walk better without tir-

(continued on page 11)

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.

Letters - cont'd from page 11

ing easily and am able to stand without holding onto something. I carry a lightweight cane not so much for walking as for standing.

The braces work great with good quality ankle high lace-up shoes...not so good with oxfords because of the rear brace adjusting screw. It will chew up the rear trouser cuff. I also did some modification to the adjusting screw since the rubber bumper wears out in a very short time.

I have nothing but praise for Arand and Bert Brinks of Rochester Orthopedics for their competency and courtesy. I have always been pleasantly surprised at their ability to keep pace with the latest innovations.

... never give up; things are constantly changing.

During my 1993 visit to Mayo's, Dr. Peter Dyck, my doctor, said that there had been success in locating the defective chromosome causing CMT, but that there were several types and wasn't sure of my type. He suggested that I leave Mayo's a sample of blood for further DNA studies. I agreed and volunteered additional samples from my two sisters. I and one of my sisters have given samples and the other sister is working on it. Dr.Dyck says the study will take time and probably will be of no benefit to me, but may help the next generation.

I believe there are many persons with neurological problems which have been correctly diagnosed, many undiagnosed, and others that have been misdiagnosed as I first was. Of course, there are those who turn their heads hoping the whole thing will go away!

Where young children are affected, perhaps ignorance and indifference by elders are the greatest obstacles.

I'm seventy-two years of age and in otherwise good health. My advice is to never give up; things are constantly changing.

F.T. New Harmony, IN

Dear CMTA,

...I first became interested in rehabilitation as a career when I attended a Jerry Lewis summer camp as a teenager. The inspiration I received from the other campers has since lead me to a Masters degree in Counseling Psychology and over seven years of professional rehabilitation experience.

I would also like to give you a brief background of my experience with CMT. It is prevelant throughout my extended family. My parents knew I had "the family foot problem" from the time I was an infant. I inherited it from my mother as did one of my brothers and my sister. It seems I have a more severe case than either of my siblings. I did not walk until I wore supportive braces. They were removed once I developed enough strength to stand and walk on my own. Later, I'm not sure exactly when, the foot problem was identified as CMT. At age 13, I had a spinal fusion with a Harrington rod insertion due to severe scoliosis. My doctor said this may or may not be related to the CMT. Since then, I have had a muscle transfer, osteotomy, and triple arthrodesis performed on both feet.

Presently, I am ambulatory without any assistive devices, although my doctor says I may benefit from having rocker bottoms put on my shoes to help alleviate occasional problems I have with my ankles becoming stiff and painful. I have little problem walking distances, but do have some difficulty with stairs, uneven ground, maintaining my balance, and, of course, performing any activities which require a significant amount of coordination.

My husband and I are expecting our first child in late June. There was a period of my life when I questioned whether or not I wanted to have children because of the CMT. However, I recall my father asking me once if, because of all I'd been through, I would rather not have been born. This was such a preposterous thought to me since the positive aspects of my life far outweigh any of the negative aspects presented by CMT. Although having CMT as a child can be emotionally traumatic at times, I can only hope that if my child has inherited CMT he or she will also inherit my positive attitude and always be glad we chose to bring him or her into the world.

Finally, my compliments to you in the production of the CMTA newsletter. I always find it informative and interesting. Good luck to you and your staff in meeting your goals for 1994. I look forward to making my contribution as a telephone volunteer.

L.M. Baltimore, MD

(continued on p.12)

CMTA Announces Volunteer of the Year Award

Robert Daino

The CMTA is pleased to announce that the winner of the 1994 Rebecca Sand Volunteer of the Year Award is Robert A. Daino, member of the Board of Directors of the CMTA.

Bob has long been associated with this organization through the local New Jersey support group which is headed by his wife, Terri. In 1991, Bob was elected to the Board of Directors of the CMTA and has been an active volunteer for the national organization since then.

Most of the members of this organization will recognize Bob's name because he was the author and moving force behind the holiday fundraising letter which went out to our membership. It was that Herculean task which best illustrates Bob's commitment to serving the CMTA.

The CMTA commends Bob Daino's volunteer work and recognizes his tremendous efforts on behalf of the entire CMT community.

The CMTA Report is published by the Charcot-Marie-Tooth Association, a registered non-profit 501 (C)(3) health organization. The newsletter is co-edited by Karol Hitt and Pat Dreibelbis. The layout is by Chesapeake Bay Design, 1418 Ticonderoga Ave., Ridgecrest, CA 93555.

Laugh Your Stress Away

Humor is one of the best on-the-spot stress busters around. It's virtually impossible to belly laugh and feel bad at the same time. If you're caught in a situation you can't escape or change (a traffic jam, for example), then humor may be the healthiest form of temporary stress release possible.

Even when you can change the situation, humor helps. Research by Alice M. Isen, Ph.D., a psychologist at Cornell University in Ithaca, New York, shows that people who had just watched a short comedy film were better able to find creative solutions to puzzling problems than people who had either just watched a film about math or had just exercised. In other studies, Dr. Isen found that shortly after watching or experiencing comedy, people thought more clearly and were better able to "see" the consequences of a decision.

The physiological effects of a good laugh work against stress. After a slight rise in heart rate and blood pressure during the laugh itself, there's an immediate recoil; Muscles relax and blood pressure sinks below pelaugh levels, and the brain may release endorphins, the same stress reducers triggered by exercise. A hearty ha-ha-ha also provides a muscle massage for facial muscles, the diaphragm, and the abdomen. Studies show it temporarily boosts levels of immunoglobulin A, a virus-fighter found in saliva.

While our cave-dwelling ancestors were stressed by actual life-threatening situations like bumping into a woolly mammoth, times have changed. "Nowadays, stress is usually not caused by the situation itself, but by how we perceive that situation," says Allen Elkin, Ph.D., program director of Manhattan's Stress Management and Counseling Centers.

Getting a new perspective is what comedy is all about. Several philosophers and writers have pointed out that comedy and tragedy are different ways of looking at the same stressful event. Comedy works by stepping back from a situation and playing up its absurdities. The same kind of disinterested observation makes the tale of your disastrous vacation seem funny- after you get safely home. For stress busting, the trick is to find ways to laugh at the situation while it's happening. Even if you don't consider yourself much of a comedian, here are a few simple techniques:

"stress is usually not caused by the situation itself, but by how we perceive that situation"

The Bart Simpson Maneuver. How would your favorite cartoon character or comedian react? "Imagining what would happen can give you a chuckle, making the situation less annoying. You can even pretend you're the star of a TV comedy, and this frustrating episode is tonight's plot," says Steve Allen, Fr., M.D., an assistant professor of family medicine at SUNY Health Science Center, Syracuse (yes, he's the son of well-known comedian Steve Allen).

Ballooning. In your mind, consciously exaggerate the situation: Blow it completely out of proportion and into absurdity- into a comedy routine. In that long, long checkout line, don't say "This waiting is killing me; I hate this." Say: "I'll never get to the front of this checkout line. The woman ahead of me is covered in cobwebs. The guy in front of her grew a beard standing in line. The cashier must be part snail. The continental drift moves faster.": This maneuver helps take the edge off the situation, redirects your tension and helps you see things as not so impossible after all. Your running commentary, however, is probably best kept to yourself. If people stare because you seem to be laughing for no reason, pretend you're reading the scandal sheets. You don't have to be a master of one-liners to be funny. There are gentler forms of humor that can diffuse anxiety in a group without making anyone feel like the butt of the joke.

Reprinted from "Wrinkle City" with permission of the artist, John H. Wheeler, Waterford, NY

Laugh - cont'd from page 12

Clip a cartoon. Keep a file of jokes and cartoons that make you laugh. Paste a few up where you're likely to need them.

Pick a safe subject. Making fun of your own foibles can save face in an embarrassing situation- you'll have people laughing with you, rather than at you. Inanimate sources of frustration, like computers and copying machines, are safe objects of humor.

Lay it on the line. Sometimes just telling the truth or pointing out the obvious can get a laugh. People are accustomed to exaggeration and truth bending (too many TV commercials, perhaps), so plain speaking can come as a refreshing shock. For example, after delivering a series of lengthy explanations during a question-and-answer period, some people have been known to put everyone in stitches by simply replying to the next question with "Gee, I don't know." "This kind of humor is a way of fighting stress by accepting our shortcomings," says Joel Goodman, Ed.D., director of the HUMOR Project in Saratoga Springs, NY. Clip a cartoon. Keep a file of jokes and cartoons that make you laugh. Paste a few up where you're likely to need them.

by Steven Lally

PREVENTION, June 1991

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Letters - cont'd from page 11

Dear CMTA,

I recently found out about your publications after being fitted for some new braces. I am a 23 year old college graduate who was diagnosed with CMT at about age 18. It affects my lower legs the most. I have very little feeling in my feet and must wear braces to maintain my stability. I love sports but playing them can be very painful. I haven't run or jumped since I was young, but with understanding friends and family, I can still compete in basketball and a Nerf football game every now and then. My disability can be, at times, very hard to deal with.

After discovering your publication, I know I am not alone. I like reading the personal stories. Many of them sound so familiar. Please send me your mailings.

T.P. MN

Dear CMTA,

Thank you so much for your educating and supportive newsletter. My daughter, age 12, has recently been diagnosed with CMT. We have found the information you sent extremely helpful in understanding our daughter's condition. Even our local doctors have been impressed. We will continue to look forward to any information in the future.

Thanks again.

I.L. Victoria, KS

Dear Friends,

I wrote to you a few months ago regarding my situation with having been diagnosed with CMT Type II when I was being worked up for spinal surgery. The spinal fusion was performed on July 15th. I had read accounts of CMT people whose symptoms were exacerbated by trauma, surgery, etc. I had also read that many doctors discounted this premise, calling it an example of "anecdotal evidence." Well, here's an anecdote for you; since the operation on my spine, the weakness in my legs has gotten very noticeably worse. I've experienced numbness in my fingers, cramps in the calves and forearms, and the toes of my right foot have begun to curl up when at rest. I had no significant "down-time" following surgery. I was up and about within a few hours afterwards, and out of the hospital the 2nd day after the surgery. Therefore, the symptoms seem to be more related to the CMT, rather than the operation, which was successful.

You lovely folks at the CMTA put me in touch with a man in Illinois who also has Type II. Talking to him was almost eerie in that our experience with CMT is so similar, i.e. no major deformities, but weakness in the ankles, a slapping gait, and fatigue,fatigue, fatigue. He told me that he had gotten an AFO and that it helped a great deal. My neurologist has prescribed one for me, for the left foot only, to help with the fatigue factor and to help with balance problems. My insurance company specifically denies coverage for fitted AFO's, so it may be a bit of a problem to find one at a reasonable price. The neurologist says I should get a fitted AFO rather than an off-the-shelf model so I'll keep looking until I find the most reasonably priced maker in my area.

I'm still dealing with the emotional aspects of having this chronic condition; some of the people close to me consider CMT to be a phantom illness, an excuse for laziness, etc. I find this attitude exasperating beyond words...my wife, however, has been very supportive and wonderful in her understanding, thank goodness. She has watched me "slowing down" for several years, and now that both of us understand what's happening, we can approach things in a practical way. I'm looking into finding a second career, something less physically demanding than my present job as a piano tuner and rebuilder. We shall see!

Let me again express my gratitude for the CMTA. You have been a source of real help to me and I really appreciate your generous help.

R.M. Yucca Valley, CA

Conference Alert
June - Rochester, NY
November - New Orleans, LA

As we go to press we are working out the details of a June CMT patient/family conference in Rochester, NY.

We are also finalizing arrangements for a November CMT patient/family conference at Louisiana State University. Our host will be Dr. Carlos Garcia, and Dr. James Lupski of Baylor Medical Center will be one of the featured speakers.

Details on both conferences will be published in the next issue of the ***CMTA Report.***

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
Abby Wakefield (212) 722-8052

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

Oregon- Portland
Mary Elizabeth York (503) 246-4939

Pennsylvania– Delaware Valley
Dennis Devlin (215) 269-2600 work
(610) 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

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 - Naselle
Marlene Russell (contact person)
(206) 484-3116

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

Support Group Leader profile

Joan Plant

Joan Plant is the leader of the CMT support group in North Central West Virginia. She lives in Elkins, WV, and works as an interviewer with the West Virginia Bureau of Employment Programs. She has CMT and is a member of a very large CMT family. She can trace CMT to her great grandmother who had ten siblings and ten children. Her own grandmother had eleven children, most of whom had CMT.

She continues to work every day with the aid of an Articular AFO, but has backaches and hip problems. She has difficulty negotiating stairs without handrails. In the winter, she uses a cane to lessen the chance of falling on ice.

Joan is a volunteer with the Reach to Recovery Unit of the American Cancer Society. She has been cancer free for the last seventeen years and continues to have a yearly check-up. She has been widowed for six years and has a son and two grandsons, who show no signs of CMT.

When Joan learned that she had CMT 15 years ago, she was told that there was no known cause for CMT, no cure and no treatment. She resigned herself to being in a wheelchair someday. The turning point for her came when her family participated in a study with Dr. Jeffrey Vance and his colleagues at Duke University. This was the study which concluded that CMT Type IA is linked to Chromosome 17. She began thinking about starting a support group and was encouraged by a cousin who attended support group meetings in Colorado.

She began the West Virginia group a year and a half ago with six or eight people. The biggest thrill she has had with the group was the day that atten-

(continued on page 15)

Support Group Leader - cont'd

dance soared to twenty-five. Considering that Elkins is a small community (8500) with no large cities nearby, participants must travel on two lane highways over mountains, and some must drive as much as an hour and a half, Joan is clearly offering something that the people need and want.

Joan admits to being discouraged at times when the attendance is low, but she hangs in there. As she said, "I believe we have a solid, very supportive group of friends - not just a roomful of strangers with the same disease."

Joan would like to hear from patients or family members who are interested in her support group. You can call Joan in the evenings at 304-636-7152. §

CORRECTION

In the Fall newsletter Donald Hay's phone number was incorrectly listed. If you are interested in a Boston Area Support Group, please call Don at 617-444-1627.

CMT IN THE NEWS

The Philadelphia Inquirer published an article on the front page of its December 24th edition called, "Gene found for disorder of the nerves: Penn scientists located the cause of Charcot-Marie-Tooth disease, which impairs mobility." The article focused on the work of Kenneth Fischbeck of the University of Pennsylvania in pinpointing the gene for the form of CMT on the X chromosome.

The Philadelphia Inquirer also published an article on the CMTA in the Neighbors Section of the December 20, 1993, issue. The article quoted Karol Hitt and emphasized the effort the organization is putting into "getting the word about CMT out."

JAMA, The Journal of the American Medical Association, November 17,1993 published both the article on Drs. Phil Chance and James Lupski that is reprinted in this newsletter and an article co-authored by them and Dr. Carlos Garcia which is cited in the scientific publications article.

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)
(please send SASE)
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)
Taxol
Vincristine

Lithium and Misomidazole can be used with caution

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, 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.

..... Type IA can now be diagnosed by a blood test.

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: