

THE CMTA REPORT

Summer 1992

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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 neurological disorder. Contents © 1992, CMTA. All rights reserved.

Orthotics And The Orthotist

At some point in the life of most Charcot-Marie-Tooth patients, the consideration of an orthosis will arise. David Showers, a Certified Prosthetist-Orthotist, and the Director of the Orthotics and Prosthetics Division of the Rehabilitative Medicine Department at the Hospital of the University of Pennsylvania, strongly believes in the usefulness of a properly designed and fitted orthotic device.

According to Mr. Showers, the decision to be fitted for an orthotic device (normally a MAFO for CMT patients) will come first from the patient's neurologist or primary care physician, and he or she will write a prescription for the orthotic device. The orthotist will then confer with your physician as well as your physical therapist or occupational therapist before seeing you. Your x-rays will be examined to determine the most efficient orthotic design, if needed.



Dave Showers

When a custom-made device is determined to be the most useful for you, your certified orthotist, after getting to know you and your lifestyle, will do a plaster wrap cast of the extremity to serve as the mold for fabrication of your custom-fitted device. Your first visit to the orthotist should take approximately 45 minutes.

The orthotist will then modify your plaster mold, smoothing and adjusting it before beginning the fabrication procedure. After the modification process, the sheet plastic to be used to create the device is placed in an oven and heated to 300-450

Terms

ORTHOSIS-

- a device used to protect, support or improve the function of the parts of the body that move.
- formerly called braces, irons, stays or splints
- currently made mostly of space-age plastics

ORTHOTIST-

- the certified orthotist is a professional who designs, makes and fits orthoses.
- the designation CO (certified orthotist) or CPO (certified prosthetist and orthotist) means the person has passed a rigorous examination process by the American Board for Certification in Orthotics and Prosthetics.

MAFO-

- molded ankle-foot orthosis.

FO-

- foot orthosis-worn below the ankle, it encompasses any part of the foot; commonly, arch supports.

degrees for a specific time by the orthotic technicians. The plastic is then molded over the modified plaster mold by using a technique called "vacuum forming." The plastic is then cooled for 6 to 8 hours before it is removed from the mold. The finished orthosis will be smoothed and suspension straps will be applied. The orthosis is then ready to be fitted.

An effective orthosis which is properly designed, fabricated and fitted will aid the patient significantly in daily functions. It should be relatively easy for the patient to put on and should be both comfortable and as cosmetically pleasing as possible.

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**Major Advance
in
CMT Genetics!**
see Research Update on p. 10

MEDICAL ALERT



Certain Drugs Toxic to the Pe- ripheral 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)
Pyridoxine (Vitamin B6)
Vincristine

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

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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.

For your records, our address is:
The CMTA, 601 Upland Ave., Upland,
PA 19015 ph.215/499-7486.

If you are moving, please send us your change of address and enclose a mailing label from a previous CMTA Report.

Orthotics (cont'd from front page)

The cost of a MAFO varies from region to region, but the average cost is approximately \$ 500.00, according to Mr. Showers. The life expectancy of the orthosis will vary from patient to patient. Factors which affect durability include a person's weight, height, the activity level of the person, the extent of muscle weakness, the severity of any bone deformities, and the type of plastic utilized in fabrication.

The MAFO usually weighs only a few ounces unless it has an ankle joint or other metal component. It should not be painful to wear. Many people describe the plastic MAFO as feeling odd at first, but that sensation should pass in a short time. If the device is painful or ever becomes painful, the device should be removed and a return visit to the certified orthotist should be scheduled immediately. Necessary adjustments will be made.

When asked if the MAFO will enable a CMT patient to walk more normally, Mr. Showers offered the opinion that an orthosis is designed to either limit motion or assist motion in a particular part of the body; therefore, a MAFO should aid in minimizing an abnormal steppage gait.

Patients using MAFOs should not attempt to wear them in spiked heels, slippers, sandals or loafers. The most appropriate shoe is a laced sneaker or an oxford. MAFOs are also slippery, use-less, and dangerous without a shoe. A shoe should be worn at all times, and the device should fit well inside the shoe and not slide or move back and forth.

A sock, stocking or other garment should be worn under the MAFO. The garment will reduce friction and will help prevent perspiration which can cause irritation. Talcum powder on the leg before putting on the sock will further help keep the leg dry.

The patient should examine his skin under the orthosis each day. If the skin becomes sore or irritated, the orthosis should not be worn. Early detection of skin irritations is vital and even minor skin problems should be taken care of. Some redness of the skin under the pressure areas of the new orthosis is predictable; however, redness should disappear 10-15 minutes after the orthosis is removed. Skin that is red or irritated can be tested by pushing on the irritated skin with the thumb for five seconds. Remove your thumb and observe. If the red area turns white and then the red color returns, the pressure can be tolerated. If the red-

dened area remains red, the pressure of the MAFO is creating a potential abrasion or sore which should be seen by your certified orthotist.

The MAFO, or any other orthosis, can be cleaned with mild soap and wiped with a damp cloth. The orthosis should be allowed to dry at room temperature.

An orthosis can be repaired if it develops a small crack. However, if the breakage is severe, the strength of the MAFO would be lessened if patching were attempted. In such a case, a new orthosis would be required. The life expectancy of a MAFO averages 3 years, dependent upon factors previously mentioned.

David Showers stresses that the decision to wear an orthotic device is a serious one which must be made by informed and trained medical and technical professionals in conjunction with the CMT patient. Each patient is a unique individual and must be treated as such. Only a certified orthotist can custom design an orthosis which will be unique to the patient's needs and anatomy thus insuring the best possible fit and correction of the person's walk.

For further information on orthoses and certified orthotists, contact the American Academy of Orthotists and Prosthetists, 717 Pendleton St, Alexandria, VA 22314.

(Information for this article was provided by David Showers, C.P.O., Director of the Orthotics and Prosthetics Division of the Rehabilitation Medicine Department, Hospital of the University of Pennsylvania, Philadelphia, PA 19104.)

TYPE II CMT Patients Needed For Study

Dr. Jeffrey Vance, Duke University Medical Center, is now studying Type II CMT. Dr. Vance would like Type II families with three generations for his study. If you are a type II diagnosed family and would like to volunteer for Dr. Vance's research, please contact him at Box 2900, Duke University Medical Center, Durham, NC 27710.

(Editor's note: We strongly urge Type II families to volunteer for Dr. Vance's research. It is only with your cooperation that the work will be done.)

Patient Develops Muscle Preservation Program

by William Clarkson

According to the CMTA newsletter, CMT, "...causes degeneration of the peroneal muscles (located on the front of the leg below the knee), foot drop walking gait, foot bone abnormalities, high arches and hammer toes, problems with balance, problems with hand function, occasional lower leg and forearm muscle cramping, loss of some normal reflexes, occasional partial sight and/or hearing loss problems and scoliosis (curvature of the spine) is sometimes present." The article continues with, "...has no effective treatment, although physical therapy and moderate physical activity are beneficial."

In his helpful book, *Head First*, Norman Cousins urges the reader to not deny his diagnosis, but to try and defy the verdict. However, we also know about the apprehension of the unknown. To me, the CMT verdict seems to have a lot of unknowns. So, about two years ago, taking my guidance from Mr. Cousins, I set out to learn all I could about the facts that might contribute to the preservation of the foot muscles and the peroneal muscles in my legs (the areas where CMT affects me).

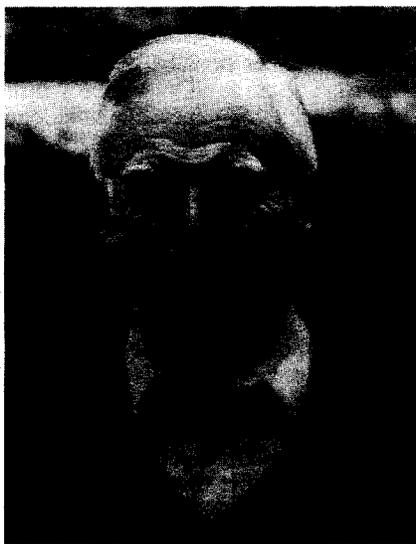
Symptoms

The symptoms of CMT for me have been a loss of feeling in both of my lower legs. The feeling loss started at the feet, progressed to the ankles, then the lower leg and in now up to the knees. This sequence started in 1975 and has continued over the following 17 years. My age is now 65.

Concurrent with the loss of feeling in the legs, there has been a decline in my ability to maintain my balance. A dramatic example of this symptom occurred when I tried to resume skating (formerly I played ice hockey for my university), after a 20 year hiatus. Balancing on the skate blades is extremely difficult, and the loss of ankle strength is no help. I am having to learn to skate all over again.

Exercise Program

As previously stated the CMTA Report says, "...physical therapy, ... and moderate physical activity are beneficial." However this statement prompts a multitude of questions. For example: 1. What type of physical therapy? 2. With what frequency, intensity and duration? 3. What results can be expected from the



William Clarkson

right combination of #1 & #2? 4. In addition to physical therapy, what other physical recreational activity might be predictably beneficial? (Activities I have found good for me are walking outdoors, walking on a treadmill, bicycling, tennis, skating, and skiing.)

With these questions in mind, I set out searching for answers. Fortunately, I was able to enlist the active and interested help of two physical therapists, Paula Machowiak and Richard Muscatello. This is a report of about two years work with Paula and Richard.

The Program

Together they recommended these exercises and this testing program:

1. Ankles, Evertors and Invertors

Daily exercises of both ankles for 6-8 minutes, using between 5 to 10 lbs. of weight on each ankle during the motions of eversion (turning out) and inversion (turning in). This exercise is designed to halt, or delay, the degeneration of the evertor muscles and invertor muscles including the peroneal muscles.

2. Ankles, Dorsi-Flexors

Daily exercise of the dorsi-flexor muscle by alternating ankle weighted exercise and standing balance exercises (five minutes each). This regimen is designed to maintain the dorsi-flexor muscles which have a major influence on foot-drop.

3. Cybex Testing

The results of these exercises are measured by means of Cybex 340 isokinetic testing at intervals of 6 - 12 months. After doing exercises #1 for 18 months the results were:

The ankle muscles (both left and right) have maintained their strength in the critical area (the eversion test) as well as the less critical area (the inversion test). One ankle lost inversion strength during one testing period. This strength was regained in 3 months by a change in the exercise pattern. The dorsi-flexor muscle program I have just begun.

Initial Observations

The strength of my ankles has been maintained for 18 months. It is important to determine if there is a direct relationship between the strength of the peroneal muscle and the ankle muscles, the latter being the ones that are being exercised. It is a great psychological benefit for the CMT patient to be engaging in a muscle preservation program with results that can be measured.

Next Steps

My action plan for the next 6 months is:

- Daily exercise following regimens # 1 & 2 and Cybex measurement of the results in 6 months.
- Continuation of daily recreational exercise, including walking (outdoor and treadmill), tennis and bicycling, and skiing and skating.

I believe that this program of exercise is helping to hold the CMT muscle weakness symptom at bay. If my report is able to be of help to any CMT colleague, I am glad.

Editor's note: The names and addresses of the Physical Therapists are Paula Machowiak, Gateway Physical Therapy, Hamburg, NY and Richard Muscatello, Sportfocus Physical Therapy, Orchard Park, NY. This article is not presented as an endorsed exercise program, but rather a course of action taken by one CMT patient. If you are interested in pursuing an exercise program, the CMTA recommends that you go to a physical therapist and follow his/her program of exercise and testing.

Funding For CMT Research

Or The Lack Thereof

It is time for the CMT community (patient, family and professional) to become actively involved in petitioning Congress to increase funding for CMT research. During 1992 grants to researchers conducting CMT research from the National Institute of Neurological Disorders and Stroke (NINDS, the division of NIH under which CMT is studied) totaled \$239,000 out of a budget of 479.3 million dollars. The total NINDS budget for 1992 was 581.1 million dollars. Obviously, CMT has not been a priority item with the government.

How can you change that...by writing to your elected Congressmen. Write your senators and your representative and request that he/she instruct the House and Senate Appropriations Committees to specifically commit funding for CMT research. A sample statement would read, "I urge you to instruct the (House) (Senate) Appropriation Committee to give priority funding for the research of Charcot-Marie-Tooth disease (CMT). CMT is the most common inherited neurological disorder affecting 125,000 Americans of all ethnic origins." Add anything else you wish but remember to sign your letter with your name and address. If you are uncertain of addresses, contact your library, newspaper, or the League of Women Voters. Any of those resources will assist you.

Our researchers are running out of money at a time when very important discoveries are being made. We must let Washington know that CMT has to be recognized and supported. The CMTA strongly urges you to advocate for yourself and the whole CMT community. If you would like to include a grey CMT brochure with your letters, send a stamped self-addressed envelope to the CMTA telling us how many brochures you need. We will send the requested quantity to you. If we all work together, this "squeaky hinge" will get some oil.

Honoraria

In honor of	By
M/M Lawrence Linker	Celia Wolfson
William Clarkson	Lilian DeArias
Sidney Weinberger	Marvin Greenwald
Ruth & Larry Linker	David Goldman
M/M Louis Solomon	Morris & Loretta Woolman
Carter J. Thompson	William H. Thompson
Michael Sand	Nancy & Joel Weisberg

Items of Interest



ITEM 1: The CMTA is most pleased to announce that Jeffrey M. Vance, MD, PhD, Duke University Medical Center, is now a member of the CMTA Medical Advisory Board. Dr. Vance's initial work on the CMT gene locus on chromosome 17 was the first to describe that location. Dr. Vance is also a clinical neurologist as well as a research geneticist.

ITEM 2: At a recent legal seminar in Philadelphia, several tidbits of interest were noted. Because of the Americans With Disabilities Act, some requirements of places of public accommodation impact on CMT patients. For instance, hotels and motels may not charge more for a room which is handicapped accessible than is charged for a similarly sized room for the general public. Restaurants must provide a employee to assist a handicapped person in any way necessary for them to be able to eat in that establishment. This means that if a CMT patient cannot open small packets, the restaurant must provide a maitre'd or server to do this task if such service is requested. Remember, you must speak up and tell public accommodation employees what you need. They cannot be expected to guess what your needs are, and, in fact, according to the new law, they may not presuppose that a handicapped person needs any special accommodations because of their disability.

Editor's note: Cards are available from the office of the CMTA which can be given to restaurant personnel requesting assistance in the opening of small packets and cream containers. Send \$1.00 and a self-addressed envelope for 10 cards.

ITEM 3: At the same legal seminar, information about job discrimination was gleaned from a meeting on the ADA (Americans with Disabilities Act). Even if you would not be covered by the new act, i.e. you work for an employer who has fewer than 15 employees, you may have legal recourse against termination of employment because your CMT has worsened and you can no longer do the job for which you were originally hired. In most states, there is long standing legislation requiring your employer to attempt to find another job for you within the firm, or to restructure your job to accommodate your needs. All of this must not place "undue hardship" on the company. If you are concerned about possible termination because of your

physical condition, please be aware that this new national legislation puts a great deal of burden on employers to be sensitive to the disabled and to make "reasonable" changes to allow the disabled to be employed or continue employment in their firm. The advice the lawyers gave was to negotiate with your company. Most companies are anxious to avoid litigation and will work with you to redefine the job to suit your needs.

ITEM 4: A database service called ABLEDATA indexes products that are available to assist with daily living activities. Up to eight pages of computer search information are provided free of charge by phoning 1-800-344-5405. ABLEDATA is funded by the National Institute on Disability and Rehabilitative Research (NIDRR) and is located at Newington's Children's Hospital, 181 E. Cedar St., Newington, CT 06111.

ITEM 5: Sue Turner, DPM, and Maggie Malone, a CMT parent, have collaborated to produce the transcript of the CMT regional conference held in San Francisco in the spring of 1991. This transcript includes the presentations of neurologist, Dr. Robert Miller, physical therapist, Dr. Frederick Bost, physical therapist, Michelle Mendoza, and occupational therapist, Sandra Cicuttin. In all, the transcription is 25 pages and is available for purchase from the CMTA for \$ 5. See the "order form" box to order the transcript.

ITEM 6: Many corporations offer to match the contributions made by their employees to charitable organizations. If you are making a donation at this time, or if you have made one previously this year, ask if your employer has a matching gift program which will double the value of your gift.

ITEM 7: At the annual meeting of the CMTA Board of Directors Diana Eline, West New York, NJ and Gary Griffith, Ocean City, NJ were elected to the Board. Diana is a research scientist and Gary is an attorney. We thank them and welcome their talents, interest and enthusiasm to the CMTA Board. The officers of the Board are Karol Hitt, President, J. Rodman Steele, Vice-President, Diane Freaney, Treasurer, Robert Daino, Recording Secretary and Donald J. Perella, Corresponding Secretary.

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, a growing and vital part of the CMTA program, inform and support anyone who must deal with this often overlooked disorder.

There are already several CMTA support groups. These chapters are spirited and growing stronger, but more groups are needed in other parts of the United States. The CMTA will gladly help you to set up a chapter in your area. For information please contact the CMTA by mail or call (215) 499-7486.

Perhaps there is a chapter 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 - Orlando Area
Carole Wray (407) 788-7427
Meets 3rd Sat every other month
Metro West Church of the Nazarene
Orlando, FL

Florida - South (Atlantic Coast)
George Meyerson (305) 431-3979

Georgia - Greater Atlanta Area,
Molly Howard (404) 333-0597
or (404) 564-9433

Georgia - Western
Molly Howard (404) 304-1652

Indiana - Indianapolis
Elaine Donhoffner (317) 841-0241
Robert Birdwell (317) 352-0235

Massachusetts - Boston,
Eunice Coben (617) 894-9510

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

Michigan - Detroit
Suzanne Tarpinian (313) 883-1123



Greg Stilwell

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

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 Muhlrig (609) 327-4392

New York City - Manhattan
Anne Beyer (201) 391-4624

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 Salzburg (919) 967-3118
(919) 286-0411 (x6586) days
Durham Medical Center

Ohio - Cleveland
Norma Markowitz (216) 247-8785

Pennsylvania - Delaware Valley
Rex Morgan, Jr. (215) 672-4169

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

Utah - Salt Lake City
Marlene Russell (801) 942-8642

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

Virginia - Richmond Area
Dennis Breckenmaker (814) 748-9021
Steve Firestone (804) 745-4123

support group corner

J. Gregory Stilwell, D.P.M., P.C., is a support group leader in Denver, Colorado. He is a podiatrist, a CMT patient and a member of a large CMT family. To say that he is very interested in the disorder is to understate the issue considerably.

In the past year, Greg, at the urging of co-leader, Margie Cowan, has hosted two CMT conferences in Colorado Springs. Each conference was attended by over 90 persons who heard presentations on genetics, neurology, and foot and ankle deformities.

Greg says of his career choice, "Because of my family history of CMT with my mother, brother, and uncle all having CMT, podiatry was a natural for me to pursue as a career." Greg is currently putting together the statistical findings of the MRI (Magnetic Resonance Imaging) results showing which muscles in the calf are typically atrophied by CMT. He uses the MRI as a tool for preoperative planning of tendon transfers and reconstructive foot and ankle surgery on CMT patients.

In the fall, Greg will go to Brazil to study with an orthopedic surgeon there who specializes in neuromuscular diseases and clubfoot. His goal is to be well versed in all CMT foot and ankle procedures and to teach in this country and internationally. Greg states his philosophy in treating CMT feet as "early surgical intervention with tendon transfers or soft tissue releases to prevent rigid bony deformity later."

Greg is married and has two sons, aged six and two. Greg believes the older one probably has CMT because of his gait and foot structure. Greg advocates, and practices, exercises of a non-weight-bearing type, such as weight training, bike riding, and swimming.

Greg is currently working on several articles to be published in professional journals which will focus on how lengthening the Achilles tendon in a CMT foot can actually make the foot worse, and how the use of the MRI can improve preoperative planning.

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.



Ask the Doctor



A patient from NJ poses several questions for the Doctor:

Are there known reasons for a CMT patient to quickly become worse?

Yes, there are several known factors which can cause an acceleration of the neuropathy. For example, CMT patients appear to be more susceptible to the neurotoxic effect of drugs such as those used in the treatment of cancer, and these may accelerate the course of the neuropathy. CMT patients may also have an increased incidence of the other (non-hereditary) neuropathies which may make their primary neuropathy (CMT) worse.

What is the incidence of CMT patients under the age of 40 who are severely affected?

We can not give an exact number, but it is very small. After 20 years of practice, I have had less than five patients out of several hundred patients wheelchair bound under 40.

Is proximal (upper arms & legs) weakness common in CMT?

Significant proximal weakness is very unusual.

Another patient asks the doctor:

Members of my extended family live in South America and seem to be less afflicted than I? Does a warm climate inhibit the manifestation of CMT?

Patients with CMT frequently suffer in the cold. However, cold does not make the disease worse it only makes some of the symptoms worse, particularly pain. Conversely, a warm climate does not inhibit the development of the symptoms of CMT.

Many patients have written to the office telling of the various diseases they were originally diagnosed with before the diagnosis of CMT was confirmed. The issue of misdiagnosis is, therefore, one we are concerned with at the CMTA. Why is CMT so difficult to diagnose?

The doctor replies:

Misdiagnosis is primarily due to a lack of awareness of the clinical manifestations of CMT. The Medical Advisory Board (MAB) of the CMTA is currently writing a physician's handbook about

CMT for distribution to primary care physicians. (Editor's note: We will announce the availability of this publication in a future issue of the CMTA Report.)

Dear Doctor:

How does CMT affect the respiratory system and what is the incidence of this? P.W., CA

The doctor replies:

CMT does involve the phrenic nerve, and as a result there will be some weakness of the respiratory muscles. Fortunately, it is very rare for this to be clinically significant.

A patient in California asks:

Can sexual dysfunction be a result of CMT?

The doctor answers:

Studies of the autonomic nervous system and CMT show some autonomic involvement. Since sexual function is controlled by the autonomic nervous system, sexual dysfunction could be associated with CMT. However no specific studies of sexual function in CMT have been done. (Editor's note: We also corresponded with Dr. Charles Brendler of the James Buchanan Brady Urological Institute, Johns Hopkins Medical Center, Baltimore, MD 21205. Dr. Buchanan also confirmed the lack of studies and he further stated he would be glad to evaluate CMT patients. Dr. Brendler may be reached at the above mentioned clinic or by calling 410-550-2329.)

Dear Doctor,

Studies on pairs of identical twins with CMT Type I have shown differences in the severity of the clinical expression of CMT I. This lends support to the concept that environmental factors may play a role in the severity. Please comment on what environmental factors would effect or lessen the severity.

The doctor replies:

One of the curious features of CMT Type I is that marked differences exist in the severity of disease within families and even between identical twins. Despite this clinical variability, there is striking

congruity between twins for nerve conduction velocity. This intertwin variability in clinical expression of disease has led to the hypothesis that environmental factors influence the clinical severity of disease. However, no consistent environmental factor has ever been identified. It is known, that increased susceptibility to nerve compression and increased susceptibility to the neurotoxic effects of drugs both occur in patients with CMT Type I. I have also seen a number of patients in whom the development of diabetes has resulted in more rapid deterioration of their neuropathy. However, in our study of two pairs of identical twins with CMT and differing clinical expression, we could identify no factor which accounted for this difference. Obviously, more work is needed on the interaction between hereditary and environment in the production of clinical deficits in patients with inherited disease.

Support Group Success Stories

Bill Porter, the leader of the Tennessee Valley Support Group, reports that 24 people attended his first meeting in May. Some people traveled from more than 6 hours distance to be part of the group. The neurologist who spoke stayed after her presentation to answer each attendee's question. It was an excellent first meeting for this new group.

Kay Flynn, leader of the Westchester County, NY, group, had her members mail 300 gray CMT brochures to neurologists, orthopedists, and other related doctors in their geographical area. This group has undertaken a vigorous campaign to increase awareness of CMT and this organization.

Rebecca Sand, a member of the Delaware Valley Support Group, has done a mailing to approximately 100 members and friends in the Southeastern PA region promoting a "no bake" sale to raise money for CMT research. To date, her efforts have raised \$ 225 for the research fund.

EDITOR'S NOTE: We like to brag about the good work of our volunteers. If you or your support group are doing CMT work in the community or have a particular success to report, please drop a note to the office and we will be pleased to mention it in the next newsletter.

CMT Patient Advocates for Health Insurance Reform

Billie Angelopulos of Springfield, IL, wrote to the organization in April in response to a paragraph in the Winter 1992 newsletter calling for cases of health insurance discrimination.

She changed jobs in May 1991, moving from a very large company to one that employs only 9 people. Her new employer provides medical insurance to employees and their dependents as a company benefit.

When she filed out the insurance application, listing CMT under preexisting conditions, she was denied coverage. She requested that they insure her for everything except the CMT, but was told they would not provide insurance for her in any form.

With her permission, Billie's letter was sent to President Bush, Congressman Curt Weldon, who represents the district in which the office of the CMTA is located, Congressman Henry Waxman who is the Chairman of the Subcommittee on Health and Environment, Pennsylvania Senators Specter and Wolford, and the *Wall Street Journal*. Further, her letter was sent on to Dr. Paul Billings at the California Pacific Medical Center, who is investigating insurance discrimination because of genetic disorders.

Billie also approached her representative, Congressman Dick Durbin of Illinois to present the health care issue at a gathering at Southern Illinois University School of Medicine. The following excerpt from Congressman Durbin's newsletter reveals Billie's success in being heard.

" Billie Angelopulos of Springfield knows what it's like to be refused health coverage because of a preexisting health condition. Billie has Charcot-Marie-Tooth disease...Last year she underwent surgery which eased her pain and allowed her to walk without braces.

At the time of surgery, Billie was working for a large company and had health insurance. Since then, she has left the large company and gone to work for a small firm whose insurance carrier refuses to cover her.

Even though the surgery was successful, the disease is not life threatening, and she is in otherwise good health, Billie still was denied health insurance because of her preexisting condition.

According to one study, as many as 81 million Americans under age 65 have medical conditions that health insurance companies might use as a reason to deny coverage or raise premiums. Millions of Americans are afraid to change jobs because they don't know whether they would get health insurance through a new employer.

I am working on a bill which I will introduce soon, which would prohibit group health insurance from excluding new members based on preexisting conditions. Those who have had a condition during the three months prior to enrollment and did not have health insurance during that time, would be subject to a six month waiting period before the ban would take effect. My bill would also prohibit rate variations, termination, and the refusal to insure or renew insurance based on employee health status or past claims experience.

Health insurance companies have not always discriminated against people with preexisting conditions. Things started to change in the 1970's when many health plans started offering lower rates to younger, healthier groups because their health care costs were lower. This resulted in higher rates for older, less healthy groups.

The bill I am drafting will impose a tax penalty on insurance companies which do not comply with the law.

This important legislation could help millions of Americans who have been denied health insurance coverage and are struggling to keep up with health care costs on their own."

If you are also a victim of insurance discrimination, or if you simply wish to make your feelings on this subject known, you can get the names of your elected officials from the League of Women Voters in your area. They have a publication called the "Legislative Directory" which they will make available to you at no cost. The CMTA urges its members to be advocates of health care reform and to contact their representatives regarding this very important current issue.

Help With Shoes Available

The following pedorthists can assist you in fitting prescription footwear or in finding you shoes that will be comfortable for the CMT foot.

Louie Winskowski
Northwestern Artificial Limb & Brace
510-8 25th Ave North
St Cloud, MN 56303

Robert Schwartz
Eneslow Comfort Shoes
924 Broadway
New York, NY 10010

Marvin Kinne
Kinne Shoe Repair
238 James St.
Utica, NY 13501

Ernest Rueda, Jr
Turnpike Orthopedic Shoes
186-06 Union Turnpike
Flushing, NY 11366

Fred Toenges
F.W. Toenges & Sons
2415 Hobson Rd
Ft. Wayne, In 46805

Mead Alaimo
Acor Orthopaedic, Inc.
18530 S. Miles Parkway
Cleveland, OH 44128

Martin Bacon
Van Dyke & Bacon, Inc.
5851 York Rd
Baltimore, MD 21212

Stewart Crenshaw
Marshfield Clinic
1000 N. Oak Avenue
Marshfield, WI 54449

Steven Thorburn
A.R. Menzies & Sons
33 Rookwood Ave.
Fredercton, New Brunswick,
Canada E3B2L8

Harriet Cavanah Dart
Dart/Ullman Prescription Footwear
674 Milwaukee Ave.
Prospect Hts., IL 60070



Memorials

In memory of
Joann M. Trautwein
Robert Barg
Ida Gutter
Beatrice Zornberg
"Beloved John"
DeChristino
D.K. Bernard

Benton Bohannon
Bernard Londner
Edward Scarsella

By
Margery Hankinson
Monica Barg
Marvin Greenwald
Marvin Greenwald

Marvin Greenwald
David &
Diane Buckman
Shirley Hubbard
Rita Londner
Flora Motta

CONFERENCES

Past...

In April Dr. Walter Bradley, chairman of the Department of Neurology at the University of Miami Medical Center, and the CMTA held a CMT patient/family conference at the Miami Medical Center. Dr. Bradley, an internationally recognized CMT authority, and his distinguished colleagues educated and enlightened the assembled CMT patients, families and friends. A group of conference attendees are forming a support group for Southern Florida. For information about this group call the CMTA office at 215/499-7486.

Future...

Chicago, IL will be the site of the next CMTA conference. The meeting will be at 10 AM, Saturday September 26, 1992 at the University of Chicago Medical Center in the Doctors' Dining Room which is located on the second floor of the Old Billings Hospital. (Directions will be sent upon registration) Dr. Barry Arnason, chairman of the Department of Neurology at the University of Chicago Medical Center and a member of the CMTA Medical Advisory Board, will lead off the conference speaking about the neurology of CMT. The second presentation of the morning will be "CMT in Children". Following lunch presentations will be given on "The Genetics of CMT" and "Surgical and Rehabilitative Approaches to CMT". Included in each speaker's presentation will be time for questions from the audience.

If you need overnight accommodations, a block of rooms is being reserved in the nearby Ramada/Howard Johnson complex. This block will be held until September 9th, and more information can be obtained by calling 800-237-4933. If stairs are a problem, request a first floor room or a room in the building with an elevator. The hotel has a free hourly shuttle service to the Medical Center. For those needing transportation from O'Hare Airport there is a reasonably priced shuttle service. The fee for the conference is \$20.00 and includes lunch. Reservations must be in by Friday September 18, 1992. Late reservations will be accepted, but lunch will not be included for reservations arriving after the 18th. We must notify the caterers on the 18th.

We encourage you to attend and learn about CMT, but also attend to meet with others with whom you share a common concern.

Referrals Available From The CMTA

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 practitioner 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 envelope indicating the geographic areas needed to: CMTA, Crozer Mills Enterprise Center, 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, phone 303-755-4588.



Three volunteers from the Delaware Valley Support Group (PA) were honored at a recent luncheon meeting. Recipients of certificates of appreciation for their generous help in completing membership mailings were Ella Martin, Dennis Devlin and Theresa Donikowski.

LETTERS

We want to hear from YOU!

Write us at:

Letters / The CMTA
Crozer Mills Enterprise Center
601 Upland Avenue
Upland, PA 19015

Chicago CMT Conference Registration

name(s) of attendees _____

address _____

telephone number _____

Fee for one person \$20.00

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

Number of persons attending _____

Amount enclosed \$ _____

Return by Return by Friday September 18, 1992 to : Pat Dreibelbis
CMTA
601 Upland Ave
Upland, PA 19015

Letters...



to the editor

Dear CMT Center,

I noticed in the newsletter that various patients had contributed comments, especially pertaining to problems with mobility.

I spent a full 8 hour day in Dallas in 1981 with a doctor who does not practice medicine, but has done research on CMT for many years. He was absolutely amazed when I saw him, that I could walk upright, that my feet lined up properly and that I was not on pain medication. This amazement came after he had tested me and determined the extent to which CMT had advanced in my body.

I do suffer a very high degree of pain in my feet. There is no natural feeling in my feet and legs from my knees down. When I press my foot against the brake pedal in my car, it is exactly as though my knees are receiving the pressure.

Yet, I walk straight, without the aid of an instrument; I drive; I work in my garden. I appear normal to observers.

I "trained" myself to walk straight. I "trained" my feet to not shift or flop or turn over. I learned how to balance those feet which had no feeling. The doctor called it a form of self-hypnosis which I practiced as a young man.

CMT was diagnosed 8 years ago. The first diagnosis was Lou Gehrig's disease-my father's killer.

CMT has affected my feet and legs, my hands, my shoulders, but it does not prevent me from any activity I choose to participate in. Of course, my activities are somewhat limited by my 71 years of age.



Do you know 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.

Oh, I would encourage CMT patients to fight back! My case has been diagnosed as "extremely severe", yet I function 98% normal.

I do suffer from a very high degree of pain. I found that putting a lot of weight on my feet at night I can sleep sometimes for 3-4 hour stretches. I fold quilts and pile them on my feet. It helps.

One of my sons-in-law is a medical doctor. For years I have refused to take any narcotic for pain. Last October, I was convinced to try Percocet. I restrict myself to 2 tablets, twice a day. It is occasionally tempting to take more, but I resist the temptation.

R.B.M.

Dear Editor,

Thank you for my first exposure to the CMTA REPORT. I want to find out as much as possible about CMT. Currently, I am seeking information and trying to seek out others who have been diagnosed with this disease and make Dr. Lupski's work known to those people.

I am currently communicating on one of those computer networks (Prodigy) and have had responses from 3 women whose husbands have been diagnosed with CMT and one lady whose daughter has it. I have given your address for all those who may be interested. These people are from all over the US.

At my next doctor's appointment, I plan to introduce this literature to my doctor and request that he provide your address to any patient he diagnoses with CMT.

Thank You.
C.D. Michigan

Dear Editor,

I am a CMT patient and I have enormous neuro pain (a deep dull aching pain) in my anterior thighs. I have found that I can decrease the pain by sleeping in a confined, straight position using foot drop splints, forearm splints and hand splints. I am interested in corresponding with other CMT patients who experience neuro pain.

If you would be interested in corresponding with me, please write me :

Elvira Bachovchin
c/o CMTA
601 Upland Ave
Upland, PA 19015

Dear CMTA,

Thank you for the information you sent me last week. I am not sure whether I have CMT or not. If not, I surely have its first cousin. The doctor at Johns Hopkins calls it a peripheral neuropathy that may be CMT or not.

I am 48 years old. Both I and my father have very high arches and hammer toes. For him, it seems to stop there. For me, it does not. About 1979 or 1980, I began to have problems with my hands...losing my grip...my left wrist would buckle when I tried to squeeze a toothpaste tube left handed. Things continued to, very slowly, get worse, and I went through a series of doctors who had a series of ideas. Finally, in 1985, I was told I had ALS. My local doctor said he did not accept that diagnosis, and he made arrangements for me to go to Johns Hopkins. They did a lot of tests, including a nerve and muscle biopsy, and decided that I definitely did not have ALS. I had a peripheral neuropathy which they have been trying to define ever since. I don't think they have done any actual genetic test. As things now stand, I have:

- very thin bony feet and lower legs with very high arches and hammer toes.
- muscle degeneration in the right hand, although the hand works okay except for the little finger.
- mild curvature of the spine which I was born with.
- loss of function in the muscles that raise & extend the left wrist & fingers.
- muscle cramps, pain, and fatigue on occasion, chiefly toward the end of the week. By Thursday and Friday, I am a less than truly productive worker.
- cold and extreme heat seem to effect me. I do best in spring and far worse in the winter.

I am lucky in several ways. I have a good job with the Federal Government where I am essentially protected by law and where I am guaranteed health insurance. For exercise, I have a small sail boat which is my "tree house" -a place I can retreat to. I do not have much of a social life because very few single women want much to do with a person who is not quite normal. But things could be worse...I could have had ALS.

Thanks again.
B.H. Virginia



Significant CMT Gene Finding

The research groups directed by Drs. James Lupski and Pragna Patel at Baylor College of Medicine initiated a major effort to identify

the gene responsible for autosomal dominant CMT. In collaboration with Dr. Carlos Garcia, Louisiana State University, large families which had a history of this form of CMT were identified and blood samples collected from as many available affected and unaffected family members amounting to a total of over 600 people. The genetic material (DNA) was purified from the white cells in the blood. These DNA samples were used to track the position of the gene responsible for causing CMT. Initial results mapped the gene for CMT in these families to the short arm of chromosome 17. On the road to refining the position of the gene on chromosome 17 in the laboratories of Drs. Lupski and Patel, a surprising observation was made. It was noted that CMT patients had three copies, rather than the usual two, of DNA sequences of a portion of chromosome 17. This led to the hypothesis that CMT

was caused by a duplication of DNA sequences. This finding has generated much excitement among human geneticists because it is unprecedented and could have implications for other genetic disease. It was surprising to find that this same mutation was observed in a number of unrelated patients of different ethnic origin.

Recently, a patient with a large duplication of chromosome 17 which was actually visible under the microscope, was identified and in a study led by Dr. Lupski, the hypothesis that CMT was due to a gene dosage effect was proven. This finding which was published in the April 1992 issue of *Nature Genetics*, may have important implications for therapeutic strategies for CMT1A in the long term because it suggests that it may be possible to correct the defect in CMT1A patients by developing methods that modulate expression of the gene(s) which map within the duplication.

A human gene encoding a peripheral myelin protein, PMP22, was recently cloned and mapped within the CMT1A duplication in a further study led by Drs.

Lupski and Patel at Baylor and by Drs. Ueli Suter and Eric Shooter at Stanford University. Earlier this year, the mouse Pmp-22 gene was shown by the Stanford group to have a point mutation in a mutant mouse called Trembler, which has many similarities to the human disease CMT1A. The PMP22 gene is expressed at high levels in Schwann cells which wrap around the peripheral nerves and play an important role in the synthesis of the nerve insulating protein myelin. The studies on the human PMP22 gene, published in the June 1 issue of *Nature Genetics*, suggest that the PMP22 gene when present in three copies instead of the normal two copies, may lead to the CMT1A condition. While there are likely to be other genes within the CMT1A duplication region, the findings of a point mutation within the Pmp-22 gene in the Trembler mouse strongly suggest that the PMP22 gene is likely to play a very important role in CMT1A and that normalization of the expression of this gene in CMT1A patients who have the duplication may be a possible therapeutic strategy to target in the long term.

RESEARCH UPDATE

by Roger Lebo, Ph.D.

Four speakers at the CMTA's Medical Advisory Board (MAB) meeting summarized the recent scientific advances in CMT research. This information had been presented at three recent meetings in the last two months. The meetings were: Chromosome 17 Workshop, Park City, Utah; CMTA MAB Meeting at the American Academy of Neurology, San Diego, CA; and the Peripheral Neuropathy Association Meeting, Rapallo, Italy.

Previously, the most common form of CMT1, slow nerve conduction CMT, was mapped to chromosome 17 (CMT1A) by Dr. Jeffrey Vance, Duke University. Other types of CMT have been mapped to chromosome 1 (CMT1B), chromosome X (CMTX), and another chromosome (CMT1C). CMT2, normal nerve conduction CMT, has not been mapped.

Dr. Phillip Chance, University of Utah, summarized the CMT1A results. Previously Dr. James Lupski, Baylor College of Medicine, Houston, TX, found that an extra copy of the CMT1A gene region resulted in the disease. Now Dr.

Chance and Dr. Lupski have each concluded that patients with three normal copies of the chromosome arm carrying the CMT1A gene cause the disease. In contrast, people without CMT carry two copies of this gene. These patients also have a number of additional problems because many other normal genes are found in extra copies in these patients. Dr. Lebo independently came to this conclusion because at least three different size duplications were found in unrelated CMT1A patients with the same symptoms. The author's conclusion was not complicated by the presence of many other extra genes causing other problems.

The chromosome region carrying the CMT1A gene has been localized to about 1% of chromosome 17 by Dr. Christine van Broeckhoven's group in Belgium. Other groups have all confirmed this work and agree on a composite map of the available probes. Dr. Chance has isolated the entire human CMT1A gene region carried in large segments attached to different yeast chromosome segments that grow in laboratory strains of yeast. Drs. Chance, Vance, Fischbeck and Lup-

ski have begun testing these clones to find genes in the CMT1A region. Dr. Chance is also searching for the CMT1C gene.

Dr. Kenneth Fischbeck, University of Pennsylvania, spoke about his work on the large isolated CMT1A gene region segments that led to a map of the shared probes. He then described the studies designed to find the CMTX gene resulting from a consortium including Dr. Victor Ionasescu, University of Iowa, and Dr. Trofatter, Harvard University. Eight large families with CMTX affecting women carrying the CMT gene have confirmed the previously reported gene location and can be tested with markers that are closer to the CMTX gene.

Dr. Jeffrey Vance spoke about his research for the CMT2 gene as well as his CMT1A results. (See the Call for Patients on pg. 2) Gene searches usually take several man/woman years in the laboratory. Continually improving probes made available by all scientists studying human genes has increased the certainty of finding any unknown gene to greater than 70 %.

Dr. Lebo Survey Results

The CMTA survey (June '91, Dr. Roger Lebo) to which many of you responded found that 75% of CMT families are interested in a presymptomatic or prenatal test if one becomes available. The author of the survey, Dr. Roger Lebo, University of California San Francisco, spoke about a prenatal and presymptomatic test for CMT1 his team has been developing. Dr. Lebo has been serving on a DNA Diagnostic Committee, Pacific Southwest Genetics Network, that distinguished a clinical prenatal test from a research test as one that is informative in at least 7 of 10 pregnancies and is at least 95% reliable. In order to be informative in at least 70% of CMT1 families, both CMT1A and CMT1B must be tested.

Previously, Dr. Lebo developed a new procedure to identify a disease prenatally for a missing (deleted) gene that sees one control red spot and one green spot on each normal chromosome. A chromosome with a missing (deleted) gene has no green spot. Dr. Lupski first used this same procedure to identify CMT1A chromosomes with one red spot and two green spots because the gene is duplicated. Unlike other DNA tests, Dr. Lebo's test can diagnose an unborn fetus with CMT1A within one week. The same test can be used as easily on children and adults at-risk for CMT1A who have not yet shown any signs of the disease. This test looks reliable based upon the results for the first forty subjects. Test of more subjects are expected to continue to confirm the reliability. Although facilities for a presymptomatic test are in the planning stages, Dr. Lebo is currently accepting requests for a research prenatal test.

Answers to another survey question indicated that a small but real fraction of women experienced a significant decrease in muscle function during pregnancy. Although Dr. Peter Dyck has not found this in his extensive practice, Dr. Robert Lovelace, Columbia University, New York City, has found significant evidence that some women have severe problems. Currently Dr. Lovelace is studying patient histories to learn more about patient problems during pregnancy and following an accident. Should a woman notice severe difficulties developing during pregnancy, Dr. Lovelace suggests that you consult your neurologist and/or call him at (212)305-2121 or (212)305-1328.

CMTA Membership/Order Form

Name: _____

Address: _____

Phone Number: _____

Tell us about yourself:

- | | |
|---|---|
| <input type="checkbox"/> CMT Patient | <input type="checkbox"/> Medical Professional |
| <input type="checkbox"/> Interested Supporter | <input type="checkbox"/> CMT Family Member |
| <input type="checkbox"/> Other | |

Enclosed is:

- | | |
|--------------------------------|--------------------------------|
| <input type="checkbox"/> \$25 | <input type="checkbox"/> \$50 |
| <input type="checkbox"/> \$100 | <input type="checkbox"/> 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)
- Booklet - CMT FACTS (\$3)
- Transcript - San Francisco CMT Conference (\$5)
- Letter - to Medical Professionals regarding the drug list (free to members with self addressed stamped business envelope)
- List - Physician Referrals (by state) (free to members with self addressed stamped business envelope) please list states: _____
- Medical Brochure - CMT (gray brochure) (free to members with self addressed stamped business envelope)

Total amount enclosed: _____

Contributions are tax deductible.

Please make checks payable to the CMTA.

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.

Peripheral Neuropathy Association Meeting

Dr. Peter James Dyck, Mayo Clinic, Rochester, MN, who organized the current and previous Peripheral Neuropathy Association meetings, called a special session in Rapallo, Italy to discuss ethical issues. Previously, prenatal diagnosis has been offered for only about twenty life-threatening genetic diseases. Dr. Dyck asked whether patients who had insurance companies pay for other genetic tests might suffer an increase in rates or be denied insurance in the future. A discussion about genetic counseling between neurologists and geneticists emphasized that geneticists strive to give the patient the results and explain the significance and risks of those results. However, counselors then ask the patient to decide what if any option to take. This differs from routine neurological practice in that the physician recommends the best treatment based upon his larger experience and then proceeds.

Editor's Note: The CMTA will announce in the CMTA Report when the Chromosome 17 diagnostic test is commercially available.

1992 Poster Child



The CMTA is pleased to announce that Sarah Nelson Collier of Baltimore, Maryland has been chosen to represent the Association as our poster child from June 1992 to June 1993.

Sarah is 9 1/2 years old and is the daughter of Ivi and Jeff Collier. She has a 21 year old sister, Laura Forsythe. Sarah was diagnosed with CMT at the age of 5 and has a mother and grandfather with the disorder.

Among her interests, Sarah lists reading, bowling, swimming, acting, singing in the school choir, and participating in community service activities. She is an "A" student and is a peer helper in her school.

Sarah and her mother are part of a program called DisABILITY AWARENESS Project (DAP) which strives to help students and teachers understand and accept other children with disabilities. Sarah regularly speaks to students in Howard County, Maryland and shares the story of her disorder and her enthusiasm for life.

As a result of her work with DAP and her work for the MDA and as a junior ambassador, Sarah was the 1992 recipient of the "Yes I CAN" Award.

Sarah and her family are very active in CMT research through Johns Hopkins Medical Center in Baltimore.

The CMTA is delighted to have Sarah serve as our poster child for the year 1992 - 1993 and wishes to thank the many other children (and their parents) for submitting applications.

CMT...

- is the most common inherited neurological disease, affecting approximately 125,000 Americans.
- is also known as peroneal muscular atrophy and hereditary motor sensory neuropathy.
- is slowly progressive, causing deterioration of peripheral nerves which control sensory information and muscle function of the foot/lower leg and hand/forearm.
- causes degeneration of peroneal muscles (located on the front of the leg below the knee).
- causes foot-drop walking gait, foot bone abnormalities, high arches and hammer toes, problems with balance, problems with hand function, occasional lower leg and forearm muscle cramping, loss of some normal reflexes, occasional partial sight and/or hearing loss problems and scoliosis (curvature of the spine) is sometimes present.
- does not affect life expectancy.
- has no effective treatment, although physical therapy, occupational therapy and moderate physical activity are beneficial.
- is sometimes surgically treated.
- is usually inherited in an autosomal dominant pattern, affecting half the children in a family with one CMT parent.
- may become worse if certain neurotoxic drugs are taken.
- can vary greatly in severity, even within the same family
- is the focus of significant genetic research, bringing us closer to answering the CMT enigma.

THE CMTA REPORT

*information on Charcot-Marie-Tooth disease
from the*

Charcot-Marie-Tooth Association

Crozer Mills Enterprise Center
601 Upland Avenue
Upland, PA 19015

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