Charcot-Marie-Tooth Association

THE CMTA REPORT

Summer 1991 Vol.6/No.3

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 © 1991, CMTA. All rights reserved.

CMT Poster Child Chosen

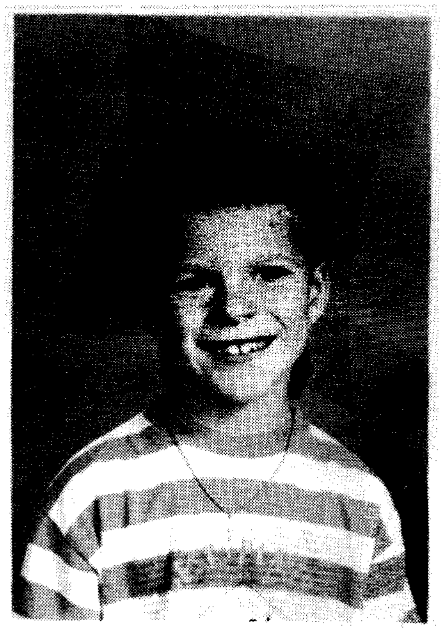
The CMTA is pleased to announce that Adam Michael Cavanaugh of Madison Heights, Michigan, has been chosen to represent the association as our poster child from June 1991 to June 1992.

Adam is 8 1/2 years old and is the son of Paul and Julie Cavanaugh. He has a brother, Alex, and a sister, Amy. He was diagnosed with CMT at age 4.

Among his interests, Adam lists Cub Scouts, basketball, bike riding, camping, collecting baseball cards, and reading.

Adam's parents describe him as " intelligent, articulate, and friendly. He has been dealing with CMT for most of his life and he doesn't let it prevent him from trying new things."

IN THIS ISSUE:



Adam Cavanaugh

Adam was chosen by three persons not affiliated with the CMTA: a freelance writer, and two company presidents.

The CMTA wishes to thank all the parents and children who took the time to submit applications. We'll be choosing another child next year, so plan to enter then.

CMTA Establishes CMT Research Fund

At the Spring meeting of the Board of Directors, the Board voted to establish a fund designated for CMT research. Furthermore, the Board contributed the initial deposit the to the fund. Any contributions now designated for research will go directly into this fund. When there are sufficient monies in the fund, the CMTA will issue a request for research proposals to the scientific community. Research proposals will be evaluated by members of the Medical Advisory Board and the Board of Directors will select the recipients.

We are very pleased that we are able to increase the scope of the CMTA to include the funding of research. We encourage any of you to help us raise funds for research. There are many things, big and small, that you can do to participate. Bake sales, car raffles, white elephant sales, walkathons, Pub nights, golf tournaments, etc. etc. are all accepted ways of raising money. Consider it, remembering that any time CMT is mentioned to the public, it educates the public about this "Mystery Disorder".

WE HAVE A NEW LOOK!

This issue is printed on white paper rather than our traditional buff color because we have received a gift of in-kind printing service from the Sun Company.

The Sun Company, of which Sun Oil and Sunoco are two well-known trade names, is headquartered in Radnor, PA, near our own national office. Sun printed the spring and summer issues of *The CMTA Report* at its print shop in Philadelphia.

PATIENT PROFILE

"START SPREADING THE NEWS"

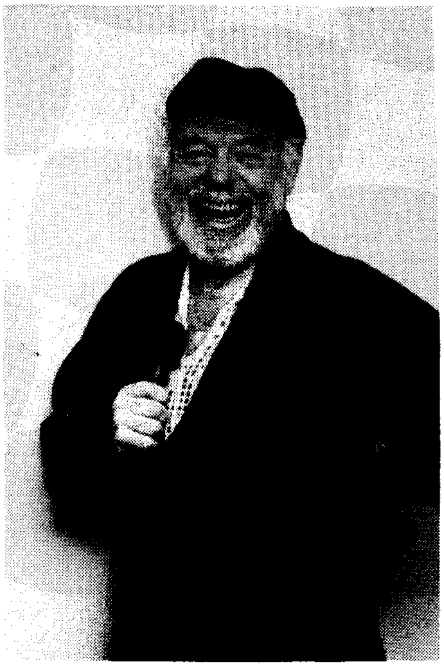
As Jack Donovan belts out his rendition of Sinatra's hit, "Start Spreading the News", he stands backed up against the piano for balance, and when he sits in his chair, the microphone stand and his legs form a tripod base that help him balance.

As an entertainer who has CMT, Jack is helping to "spread the news" about his disease and the CMTA's work in helping and educating CMT patients. He has offered to do public service announcements in the California area in which he is known.

Donovan was in his forties when his disease was diagnosed. In retrospect, he remembers not being able to climb the ropes in gym class or roller skate very well. He attributes those failings to the muscular atrophy that has since gotten even worse.

His father also had the disease but was not diagnosed until he retired from his steel forging job at 62. Donovan thinks his father's disease did not show up because of the exercise his father got constantly carrying heavy metals.

Believing that inactivity is the worst thing for a CMT patient, Jack works out as often as possible. He squeezes silly putty when he watches television and also stands on the edge of the telephone book and comes up on his toes and then lowers down onto his heels. " I'm not faithful with my exercise, but I know that when I do exercise I feel better, " observes Jack.



Jack Donovan

Jack is 61 and still actively performing American Pop and Jazz music on cruise ships and in night clubs and lobby bars in Costa Rica, Guatemala City, and Cancun.

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)
Pyridoxine (Vitamin B6)
Vincristine

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

CMTA, What It Is

The CMTA is a non-profit organization devoted to CMT disorders. It was incorporated in the Commonwealth of Pennsylvania in 1983, and is an IRS 501-(C)-(3) charity. It is governed by a Board of Directors who serve without pay. Currently, there are eleven members most of whom are either CMT patients or members of a CMT family. We are going to introduce our readership to the Board beginning this issue with an article about Rex Morgan. (See Support Group Corner)

The funding for our program comes primarily from donations from patients, their families, and their friends. We receive no governmental funding, and in light of the current economy, do not anticipate any in the near future. Our requests for funding to private foundations, so far, have resulted in one small grant.

The CMTA has one half-time employee Pat Dreibelbis. Pat is in the office twenty hours a week. All other work is done on a voluntary basis. Conservatively, at least 80 hours/week are donated by a variety of individuals. Included in this donated time is the work of our treasurer Diane Freaney. Not included in this time is all of the work done by our dedicated support group leaders.

The result of the contribution of time is that little money is spent for administration, and the major share of the money is spent on our program for the CMT community. However, our growth rate is such that we are going to need not only larger office space, but more personnel.

Our Medical Advisory Board is comprised of 42 clinical and research scientists, who serve voluntarily. We are indeed fortunate to have such an assemblage of dedicated professionals. (See the Winter CMTA Report for a list of MAB members.)

This is only a small part of the CMTA; the remaining part is you our members. If you wish to become an active part of the organization, we would welcome your participation. We need your ideas and your efforts to assist us in reaching the CMT patient and the general public. Together we can accomplish this and the ultimate defeat of CMT.

CMTA President Elected to NORD Board of Directors

In May Ann Beyer and Karol Hitt attended The National Organization for Rare Disorders (NORD) annual conference in Washington, DC. At the membership meeting Karol was elected to the Board of Directors of NORD.

The conference was an opportunity to network with other like organizations, to attend informative workshops, to learn of current NIH projects, and to keep current about legislation on Capitol Hill.

An outstanding presentation entitled "Genes and Genetics" was moderated by Dr. Jess Thoene, NORD President with presentations by Dr. W. French Anderson if NIH and Dr. Robert Nussbaum of the University of Pennsylvania.

At the conclusion of the meetings Ann and Karol visited several legislators on Capitol Hill to express concern for the reauthorization of the Orphan Drug Act. The Act authorizes money for orphan drug research, provides tax incentives to pharmaceutical companies to develop drugs for rare disorders, and gives seven-year exclusive marketing rights to the developing manufacturer. There is a movement to amend the law and put a cap on the profits of the manufacturers. The proposed cap under discussion would be $150 million dollars. Once this cap has been reached, the marketing monopoly for the drug would be lost and it would be treated like any other prescription drug subject to competitive pricing.

The Orphan Drug Act has effectively increased new drugs for rare disorders. Whereas there is no drug therapy for CMT now, certainly in the future there is a strong possibility that there will be. Therefore, it is in your interest to preserve and amend the Act. This can be done by writing to your senators and congressman and voicing your position. Local newspapers and your area League of Women Voters can furnish you with names and addresses of your legislators.

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PATIENT PROFILE

Carla McGreevey

(Editor's note: *Carla wrote to the editors of the CMTA Report with her story of CMT, and we profile her here not because her story is so unusual, but because it is so typical.)*

Before her diagnosis, Carla McGreevey was active in swimming, diving, tap dancing, gymnastics, and cheerleading. Her first symptoms appeared in high school when she began to have trouble keeping up with the gang when they walked downtown. " I thought everyone was walking faster than usual," she remembers. But, in fact, she was just walking slower and turning her ankles on tiny rocks and sticks.

In 1962 CMT was diagnosed and Carla remembers the neurologist telling her that she would never be a dancer. She cried and then went home thinking she just had to live with this disease.

She had triple arthrodeses on her right and left ankles in 1962 and 1963 and went on to graduate from college and teach kindergarten.

In 1978 she went to see what could be done orthopedically to help ease her walking which was getting more difficult. She was given a prescription for bilateral splints. " I took about ten steps wearing the splints and wanted to shout for joy,"(and did) she recalls. She has worn them since that day.

At age 39, Carla became pregnant for the first time. Worried though she was about her age and her CMT, she gave birth to a healthy baby girl who today at age 6 still has no outward signs of CMT. Currently, Carla rides bicycle, works in her yard and does virtually anything if it doesn't involve extensive walking or doing small tasks with her hands.

Carla has her own conclusion to her profile which she calls her personal "Hang-ups and heartaches."

"I am reluctant to wear skirts, except in cold weather when I can wear tall dress boots.(I always worry about what to wear to a summer wedding!) I won't go swimming in public because I have lots of trouble walking without the splints, and I am self-conscious about the muscle atrophy in my legs. I long to go roller skating and ice skating with my daughter. It hurts to rely on my husband and friends to help her at skating parties. I get tired of having such trouble getting shoes that are comfortable with the splints and that are appropriate for different occasions. Vacations can often be a chore rather than carefree fun because most involve walking long distances and unfortunately,walking is not one of my favorite things to do. And, finally, I do get tired of people who stare! My gait is not smooth, but I try to live with it and don't like being reminded of it by people who stare. In spite of all the frustrations of CMT, I can still function quite normally and I am very grateful for that!"

We need an upgrade!

Since 1986 we have been using a donated, used IBM PC XT. Due to the age of the system and the growth of the CMTA we urgently need to upgrade our system. Our computer friends tell us we need an IBM (or clone) 386 SX, 2 MB RAM, 80 MB Hard Drive, 3 1/2 and 5 1/4 floppy drives, VGA monitor and controller. If you can help us obtain this system, please contact us.

Vocational Assessment and Evaluation Options

HEATH (Higher Education and Adult Training for People with Handicaps) reports that people who want to find out about going back to school or need help in determining the appropriate direction for future job training or career changes include those who have recently become disabled; those who were disabled and employed, but have had their current job phased out; and those who are unemployed due to their lack of vocational skills training.

HEATH recommends that persons in any of these situations call the Vocational Rehabilitation Services (VR) which is the nationwide federal-state program for assisting eligible people with disabilities to define suitable employment goals and become employed. Each state capital has a central VR agency, and there are local offices throughout the state that can be found in the telephone directory under STATE listings, Vocational Rehabilitation Services. CMTA members can refer to the Winter 1990 issue of *The CMTA Report,* pages 6 and 7 for a complete state by state list of the offices.

The staff of the VR agency can provide information on the local vocational evaluation centers that are accredited and employ certified evaluators. In order to be served through vocational evaluation programs funded by VR, an individual with disabilities must meet certain eligibility requirements. For more information about eligibility requirements and the vocational rehabilitation process, ask HEATH for a free copy of "*Vocational Rehabilitation Services: A Postsecondary Student Consumer's Guide*". You can write to HEATH Resource Center, One Dupont Circle, Suite 800, Washington, DC,20036-1193. Or, you can obtain a copy of the complete article on the procedures for vocational assessment from the office of the CMTA by sending a stamped self-addressed envelope and $1 to cover printing costs to: CMTA, 600 Upland Ave., Upland, PA 19015.

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support group corner

Rex Morgan

"One of the great things about a support group is that you come away with something valuable after each meeting. It may be something simple - like the time a member showed everybody a three-sided pencil grip which made it easier for him to write - or something bigger - like strategies for coping emotionally with the disorder,"says Rex Morgan, support group leader for the Delaware Valley, PA. Rex is, himself, a CMT patient who was diagnosed in childhood.

Rex typically averages 20-25 people per meeting and plans meetings every two months at Holy Redeemer Hospital in Meadowbrook, PA. Attendees have included a lawyer, a comedy writer, a mime, a podiatrist, a former policeman, and a computer programmer, among others.

At Rex's May meeting, podiatrist Simon Small of Bala Cynwyd (a Philadelphia suburb) spoke about his company, Pro Support Systems, which manufactures a special three-layered, custom-molded orthotic. Dr. Small has as clients athletes such as Chris Everett, Larry Bird, and Joe Montana. Their foot problems (high arches, tight Achilles tendons, to name two) are not unlike CMT patients' problems. Dr. Small, therefore, addressed the needs of the CMT foot and how his orthotic could be of value to CMT patients.

Rex's next meeting is scheduled for July 13.

In addition to being the Delaware Valley support group leader since 1988, Rex Morgan serves as the Public Relations Chairperson of the Board of Directors of the CMTA. In his personal life, Rex is a professional writer and performer.

" I know that many members have gotten a better handle on their lives and their attitude toward CMT as a result of our group discussions. That's very gratifying to me. It makes all the work worthwhile," concludes Rex.

Memorials

In Memory Of	By
Eric Cooper	B. Gadsly
Ray Lawless	Mr/Mrs William Usher
Jacob J. Masel	Edythe Masel
Mrs. Pearl Safro	Mr/Mrs Henry Chow

Professional Help

The following pedorthists have become members of the CMTA and are interested in and knowledgeable about the foot problems of the CMT patient. If you visit their facility, please tell them that you found them through the CMTA newsletter.

MR KEVIN BROWN, C. Ped.
The Cobbler Shop
115 E. Superior
Alma, MI 48801

CORRECTIONS

In the Spring issue of the CMTA REPORT incorrect information was given in the article "Financial Options for the Disabled". A statement was made that after an individual has been receiving disability benefits for two years his/her spouse and/or minor dependents is eligible for Medicare. This is incorrect. After two years of receiving benefits, the disabled person is then eligible for Medicare. During that initial two year period the individual is responsible for his/her own medical coverage. Also, a surviving disabled spouse of a worker who was covered under Social Security can apply for Disability if he/she is 50 years old or older. For more information request the booklet "Disability" by contacting the Social Security office near you or call nationally, 1-800-234-5772. The national Social Security number is open from 7 AM to 7 PM on business days.

The honoraria listed for Elissa Maxwell should have read Melissa Maxwell.

The letter from S.W., Australia should have read, "I now start by doing four laps of walking with my heels down first."

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 CMT.

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.

SUPPORT GROUP LEADERS HELP SWELL THE RANKS

Three active CMTA support group leaders, Molly Howard in Atlanta, GA; David Berger in San Francisco, CA; and Greg Stilwell in Denver,Co. have recently held very well attended support group meetings or conferences and have given the names of new CMTA members to us. Their efforts have netted the CMTA a total of 70 new names in our data base. We wish to applaud their efforts and their success.

We encourage other support group leaders to publicize their meetings as widely as possible and to send an attendance sheet with names and addresses to headquarters as soon as possible.

It is our continuing goal to reach each CMT patient and, through our newsletters, tape rentals, doctor referrals, and conferences to offer each patient the help they previously were without.

Each member of the CMTA can also help by sending us the names of anyone you know who would be interested in joining the CMTA or receiving our publications. If each of us works at enlarging our membership, the goal of reaching the 125,000 projected CMT persons in the US would be achieved in no time.

If there is no support group in your area or if the support group in your area is inactive, consider getting involved yourself in the support group concept. If you are interested in leading a group, write to us at the CMTA, 600 Upland Ave, Upland, PA 19015, and we will send you the start-up information kit.

California– Los Angeles area
Burbank
Sherry Brown (818)841-8421

Oxnard-Thousand Oaks
Janice Hagadorn (805) 985-7332

San Bernadino ("Inland Empire")
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) 792-1427

California– San Francisco
David Berger (415)491-4801
after 6:00 pm

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

Connecticut
Linda Friedo (203) 374-8478

Florida– Orlando Area
Mary Beeler (407) 295-6215
Meets 3rd Saturday, every other month

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) 253-5632

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

Massachusetts– Boston,
Eunice Cohen (617) 894-9510

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

Michigan– Detroit
Suzanne Tarpinian (313) 883-1123

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 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
Meetings: Holy Redeemer Hospital
Meadowbrook, PA

Pennsylvania– Pittsburgh
Garnett McDonald (412) 937-0115

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

Ask the Doctor

We regret we can not answer all letters we receive which pose medical questions. We chose letters that ask questions which we feel would be of general interest to our readership. We have no doctors on staff at our office, and most questions are sent to physicians who serve on our Medical Advisory Board. Due to the volume of mail we receive, we are unable to respond to all letters.

Dear Doctor,

Does Prednisone seem to adversely affect CMT? I have taken it for 15 years or more.

Also, is there a possible relationship between macular degeneration and my CMT?

C.R., CA

A member of the CMTA's Medical Advisory Board replies:

In general, prednisone is not known to adversely affect CMT. It would be important to know what type of CMT you have, why you take the prednisone and how much prednisone you have been taking. Some varieties of CMT have even been reported to be improved by prednisone. However, large doses of prednisone sometimes damage muscle cells and cause a "myopathy" which can be associated with increasing weakness. You should ask these questions of a local neurologist.

As a general rule, CMT does not involve vision or the retina of the eye. However, a few rare types of hereditary neuropathy have been associated with optic nerve atrophy or retinal degeneration. Some of these cases may involve abnormalities of mitochondria, which are energy packets in the cytoplasm of cells. It would be important to know if other people in your family have CMT and/or macular degeneration. These are questions that should be referred to your local neurologist and ophthalmologist.

Dear Doctor,

My husband developed peripheral neuropathy at age 72 after he broke his hip. He now has trouble emptying his bladder (currently he is using a catheter) and defecation requires great physical effort. Are these problems a result of his CMT?

R.L., FL

The doctor replies:

This is actually a complex question and there are not enough details to provide a very specific answer. In general, CMT does not cause difficulties with emptying bladder or bowels. It would be important to know if this patient has other " associated medical problems" and whether his prostate has been evaluated. Does this man actually have CMT or some other form of peripheral neuropathy? Are there other affected family members? The patient should obtain the advice of a neurologist, physiatrist or urologist.

Dear Doctor,

I was in a very serious automobile accident in December of 1988. In December of 1989 I was diagnosed with CMT. Other than very minor problems, I had no manifestations of the disease until after my accident. I ended up with herniated discs in my neck and one in the lumbar area as a result of the accident, but I believe the accident also affected my CMT. I am in full leg braces now and have had to leave work completely. Do you know if trauma/accidents can cause CMT to appear and also to develop more rapidly than it otherwise would?

M.P., NH

The reply:

The writer asks a good question that is quite difficult to answer. It is a good question because it has never been carefully investigated in the past, and there is considerable interest in the effects of trauma and stress on all diseases. However, the question is difficult to answer because the problem includes many different factors that would be hard to control in a scientific study. For example, trauma and subsequent evaluation by physicians may simply uncover a long-standing disorder that has not really changed in any significant way. Or, the trauma may add additional damage to the body or nervous system which increases disability,but does not directly exacerbate the underlying neuropathy. Issues of unemployment, insurance coverage, litigation, and compensation may also play important roles. Of course, it is also possible that stress and trauma truly produce a direct physiological deterioration in some types of CMT. This would be an interesting study, but by no means, a simple problem to attempt to study in a scientific fashion.

Dear Doctor:

I am being treated for bipolar depression with Lithium, Deseryl, Prozac, and Navane. I had a mastectomy in 1987 and was treated with cytoxen, 5-Fu and methyltrexate for 6 months. In your opinion were the chemotherapy or psychotrophic medications toxic?

Soon I may be taking Tegretol. Will this cause any difficulty for me?

The Doctor replies:

For the most part, CMT effects are confined to the peripheral nervous system, that is in the arms and legs. Therefore, drugs which act primarily on the central nervous system should not be any more toxic to the CMT patient than to anyone else. Lithium, Deseryl, Prozac, and Navane are all drugs with a wide range of side effects, but none of these effects involve peripheral nerves,

Cytoxin, 5-Fu and methotrexate are all highly toxic drugs used in the treatment of cancer. However, none of these drugs has an effect on peripheral nerves. Therefore, it is not likely that they will have affected the progression of the CMT.

Tegretol is an anticonvulsant medication, which is also used to treat patients with bipolar depression and to treat pain in patients with neuropathy. It is not clear what the exact indication for the use of Tegretol is in this situation. Tegretol will not have a direct effect on the nerves in normal patients or patients with CMT. However, Tegretol can sometimes cause some problems with coordination and patients with CMT may be more susceptible to this side effect.

Question: (continued)

My hands concern me now. I have lost most of the intrinsics and thenar muscles. There is a burning sensation in those areas. The tendons in the palm are thickening and becoming hard. I have noted a rapid increase in deterioration in the last three years. Is this part of the CMT timetable or is it possible that writing and computer work are excellerating the process?

The Doctor answers:

The symptoms of burning sensation in the hands and increased prominence of the tendons in the palms is certainly consistent with the diagnosis of CMT. Almost all patients eventually develop weakness and wasting in the hands and the burning sensations are not uncom-

Ask the Doctor

(cont'd from pg. 6)

mon. However, it is important to recognize that CMT patients, with their enlarged peripheral nerves, are more susceptible to additional nerve damage from nerve compression. This may involve the median nerve in the carpal tunnel producing a Carpal Tunnel Syndrome on top of the CMT which could cause some of these symptoms. Careful nerve conduction studies can sometimes distinguish between the inevitable effects of CMT and the added complication of nerve compression.

Question: (continued)

I have read studies in which dementia was found in CMT patients. I have some memory loss problems. Is this coincidence or is there positive linkage?

Reply:

As mentioned earlier, the major effects of CMT are on peripheral nerves. A variety of minor problems with the brain and spinal cord have been described, but dementia cannot be considered to be related to the disease itself. It is more likely that the memory loss being experienced is a coincidental problem rather than being due to CMT.

Question: (cont'd.)

Have any other CMT patients had problems with general anesthesia? I had trouble regaining consciousness and taking deep breaths. After light anesthesia I felt fatigued, was short of breath and had an increased respiratory rate. Just recently I read that there can be phrenic nerve involvement in CMT. Are my experiences any indication of that?

L.B.,OH

The Doctor concludes:

I am not aware of CMT patients regularly having problems with anesthesia. The phrenic nerves, like every other nerve in the body, are definitely involved in CMT. However, the degree of involvement is insufficient to produce clinical problems, at least until the disease is extremely far advanced. I think that the breathlessness which followed anesthesia was probably not specifically related to CMT or to phrenic nerve involvement.

Dear Doctor:

My husband (42) and my son (16) have CMT. My son has a lot of muscle weakness in the legs and feet and has had numerous surgeries. My husband has weak ankles, but nowhere near the weakness that my son shows. Three years ago my husband had a sudden onset of scotomas in both eyes. Please, define scotomas. After seven specialists and two neuroophthalmologists, they came to the conclusion that it is optic atrophy due to CMT. They said progression is expected and there is nothing they can do to stop or slow the progression. Our question is this: Is anyone following CMT patients with optic atrophy? How bad does this condition get? We know that everyone is different and that no one can say for sure what will happen.

Thank you for any help.

J.C.

Editor's Note: This letter was answered by a consulting ophthamologist with a special interest in genetic disorders.

1. Define scotomas

The term "scotoma" is derived from the Greek word "scotos", which means "dark". A scotoma is a defect in the field of vision, typically a defect in central vision, involving or close to the point of fixation or the point of regard. Thus, "scotoma" refers to a blind or partially blind area in the field of vision.

2. Does optic atrophy occur in CMT?

CMT is the descriptive name for a group of disorders inherited in various ways. Unfortunately, because of variable severity and variable expression, the diagnosis of CMT is often made in error by physicians who do not consider alternative diagnoses or variations in other diseases with muscle weakness. Typically, the diagnosis of CMT can be sustained only after diagnostic procedures which include the demonstration of slowed nerve conduction velocities on both sides of the body, involving at least two different nerves on each side. Therefore, I wonder if the diagnosis of CMT is sustainable in this family, even if the father and son have "the same disease".

Optic nerve involvement in CMT is extremely rare. It is alleged that Charcot's original patient developed optic atrophy some 40 or 50 years after other symptoms of the disease appeared. However, optic nerve atrophy in young individuals is not at all a characteristic feature of Charcot-Marie-Tooth disease, and its appearance in a young individual is sufficiently unusual to challenge the diagnosis of CMT.

Another disorder, called the Rosenberg-Chutorian syndrome, was described in 1967. Individuals with this disorder have delayed motor development, with walking beginning at about age two or three years. Muscle weakness related to atrophy of the leg muscles occurs and gait deterioration begins between the ages of five and ten. A neurologic examination of adults shows severe peripheral weakness and atrophy of extremities, including the intrinsic muscles of the hands and feet. Individuals with this disorder appear to develop optic nerve atrophy at or after age 20. Hearing tests also demonstrate neurosensory hearing impairment. The inheritance pattern of the Rosenberg-Chutorian syndrome is debated.

3. How bad does "this condition" get?

This is a difficult question to answer in the absence of a specific diagnosis. It would further be useful to know whether the individuals affected have had MRI scan or other objective assessment of brain, visual, and hearing function. It is almost never possible to "predict" the outcome of progressive visual impairments in specific individuals. Of great concern in answering this question is the inability to predict the variability of a condition in the absence of a specific and accurate diagnosis.

CMT FACTS

AVAILABLE NOW!

CMT Facts, a 16 page informational booklet about CMT and therapies, may be purchased from the CMTA. To order complete the "*I want to be in touch!*" panel on page 11.

CMTA CONFERENCES...

Past And Future

PAST

Just under 300 CMT patients and family members attended one of three conferences in the West this Spring. The first of the series was in March in Colorado Springs and was organized by Dr. Gregory Stilwell. Dr. Stilwell, a CMT patient, a CMTA support group leader, and a practicing podiatrist, arranged an all-day program of medical professionals who spoke to an audience of over 90 people. On the following Monday morning we began receiving calls from enthusiastic attendees acknowledging Dr. Stilwell's efforts.

Dr. Roger Lebo collecting blood samples at LA conference

In April working with Dr. King Engel, neuromuscular chief at Good Samaritan Hospital in Los Angeles, an all-day conference was presented to just over 80 participants. Dr. Roger V. Lebo, a research scientist at Un. of CA, SF, not only spoke about genetics and research, but also requested CMT participants to contribute blood for his research. We are delighted to report that 23 people gave blood at the meeting, and they represented 20 families.

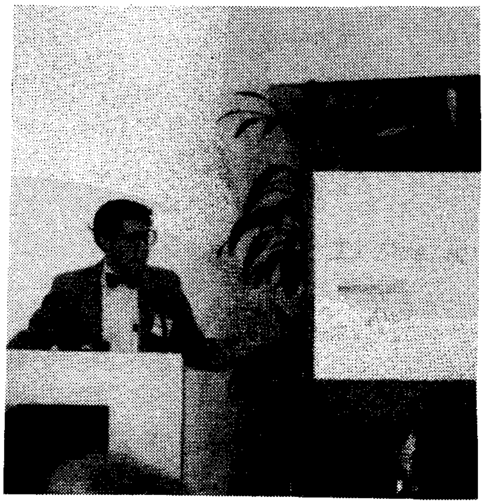
Children's Hospital in San Francisco was the site of the May conference. Dr. Robert Miller, neuromuscular chief at Children's, was our cooperating host. Just under one hundred people came to this meeting, and were well taught about CMT.

Space prevents us from reviewing each presentation at these meetings, but we would be remiss if we did not thank the hospitals and the medical personnel with whom we worked. The hospital staffs were cooperative and helpful. Dr. Engel, Dr. Miller and Dr. Stilwell gave freely their time and knowledge. We are indebted to these learned and caring professionals.

FUTURE

We are pleased to announce the **Great Lakes Area Regional Conference** will be September 14, 1991. The all-day conference, hosted by Wayne State University School of Medicine, will be in the Weber Auditorium, Harper Hospital, 3990 John R., Detroit, MI 48201. Dr. Danny Watson, Associate Professor of Neurology, Wayne State University School of Medicine, is directing the meeting.

The program will begin with registration from 9-10 AM. Beginning at 10, Dr. Watson will discuss "The Clinical Features and Diagnosis of CMT" and Dr. Orest Hurko, Associate Professor of Neurology, The Johns Hopkins University School of Medicine, will speak on the "Genetics of CMT". Following lunch the afternoon will feature presentations entitled "Orthopedic Options in CMT by Dr. Richard Lamont, Associate Professor of Surgery, Wayne State University School of Medicine; "Physical Therapy in CMT" by Steven Stewart, P.T., Children's Hospital of Michigan; and "Occupational Therapy in CMT" by Dawn Brennan, O.T.R., Children's Hospital of Michigan. The afternoon will conclude with a panel discussion by the faculty about coping with CMT. During the day each speaker will allow time for questions from the conference attendees.



Dr. Miller - San Francisco conference

Harper Hospital is easily reached by either Interstate 75 or 94. **From I 75** exit at Mack Avenue; turn right onto Mack and then proceed to John R. Turn right onto John R. and proceed 2 blocks to Harper Hospital. The parking lot is adjacent to the main entrance. **From I 94** exit at John R./Woodward. Proceed (this is a one way street) to the hospital which will be approximately one mile on your left. The parking lot is adjacent to the hospital.

For out of town attendees The Hotel St Regis is convenient to Harper Hospital and runs a shuttle service to and from the hospital. For reservations call 313-873-3000 or 800-848-4810.

Four hours of Continuing Medical Education credit will earned by attendance at the conference. For registration and information about CME contact the Continuing Education Office at 313-577-1180.

The cost of the conference is $21.00 and includes lunch. If two family members attend together the fee is $37.00 including lunch, and if a family of three attends the fee is $48.00 again including lunch.

To register for the conference complete and return the form on page 11. Early registration is encouraged, and must reach us by September 6th. We urge you to join us for a day of learning and socializing with people with like cares and concerns.

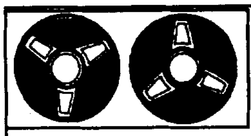
FLASH!

As we go to press, we have just learned from Dr. Greg Stilwell of the second CMT conference he is arranging. The date is Saturday, September 21, 1991 from 9AM to 4PM in the Embassy Suites Hotel, Colorado Springs, CO. Currently, the known speakers are Dr. James Drennan, an orthopaedic surgeon at Carrie Pingley Hospital, Albequerque, NM and Karen Prescott, a geneticist at Children's Hospital, Denver CO. For more information contact Dr. Stilwell at 719/594-9920.

In the planning stages for this Fall... conferences in Texas and Baltimore.

MAB Meets in Boston

The Medical Advisory Board of the CMTA met during the April meetings of the American Academy of Neurology. The meeting was chaired by Dr. Roger V. Lebo (University of California,SF). Speakers and topics were: Dr. Jeffrey M. Vance (Duke University), "The International CMT Consortium"; Dr. Phillip F. Chance (University of Utah)," Mapping the CMT1A Gene Region on Chromosome 17"; Dr. Victor Ionasescu (University of Iowa), "X-linked Recessive CMT"; Dr. Florian P. Thomas (Clinical Research Institute of Montreal, Canada), "Tomaculous Neuropathy in CMT1B"; and Dr. Kenneth Fischbeck (University of Pennsylvania), "Summary of CMT Session at Chromosome 17 Meeting". Dr. Lebo gave the concluding statements. There was also discussion about the effects of trauma on the CMT patient. This is a topic the MAB is interested in studying.



VCR Tape Rental

The CMTA has available for rental four lectures which were taped at patient conferences sponsored by the CMTA. The tapes are for play on a VHS VCR. Beta tapes are not available. The speakers are authorities in their fields and lecture topics include: Neurology, Physical Therapy, CMT Genetics, and Orthopedic Surgery.

Single lecture tapes (1 hr., 15 min.) rent for $10, and the double lecture tapes (2 hr., 30 min.) rent for $15. The rental fee includes prepaid return postage. To order a tape, fill out our "I want to be in touch!" form (see page 11) and send it to us with a check or money order payable to: The CMTA, 600 Upland Ave., Upland, PA 19015.

LETTERS

We want to hear from YOU!

Write us at:
Letters / The CMTA
600 Upland Avenue
Upland, PA 19015

CMTA Remembrances

Your gift to the CMTA can honor a living person or the memory of a friend or loved one. Acknowledgment cards sent in honor of or in memory of will be mailed by the CMTA on your behalf. These donations are a wonderful way to keep someone's memory alive or to commemorate happy occasions like birthdays and anniversaries. They also make thoughtful thank you gifts. You can participate in the memorial and honorary gift program of the CMTA by completing the form below and mailing it with your check to:
CMTA, 600 Upland Ave., Upland, PA 19015.

In honor of: (person(s) you wish to honor) ____________________
In memory of: (name of deceased) ____________________
Send acknowledge to: Name: ____________________
Address: ____________________
____

Occasion:
☐ Birthday
☐ Wedding
☐ Anniversary
☐ Memorial
☐ Holiday
☐ Thank You
☐ Other

Amount Enclosed $________

Gift given by: Name: ____________________
Address: ____________________
____

☐ Check if you would like the amount of your gift to be revealed.

The CMTA Report is published quarterly 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, 48 Henry Court,Hollywood,MD, 20636.

The opinions expressed in the newsletter are not necessarily those of the Charcot-Marie-Tooth Association. The material is presented for eduacational 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.

Honoraria

In Honor Of	By
James Thompson	Mae B. Thompson
Howard Parker	Sharon Snook
Eleene Agnew	Richeleen Snowden
Hark Wurzburg	M/M James Goldsmith
Gary P. Bogg Jr.	Gary Boggs, Sr.
Rebecca Brezel	Dr. Karen Edelson
Sophie Bradwick	Faye Bradwick
Rebecca Sand	Zahavi Fund
Hillary Bobrick	Ron Catelan

Letters... to the editor

Dear Friends,

I wish to express my appreciation to you for the conference held in San Francisco May 11th. It was extremely interesting and informative and the speakers were excellent. I am sure everyone benefited from it. I found a few things discouraging, but, overall, came away with a much more optimistic feeling about CMT and my control over it. It was by chance that I learned of the conference and your association, but I am glad that I did.

R.E. CA

Dear CMTA,

On last May 11, my wife and I attended your conference in San Francisco on CMT. Although I had been diagnosed as having CMT, this was the most helpful and informative conference on the subject that I have ever experienced.

You deserve much credit for the manner in which the conference was conducted, and we thank you for that. All of the speakers gave fine talks, and Dr. Robert Miller's presentation was indeed outstanding.

Both my sister and my neice have the CMT syndrome. My father, who was born in Indiana, is our only known ancestor to have had CMT.

We would be happy to contribute information or in any other way to assist your efforts. As for my part, future donations will go to the CMTA to help further your great efforts.

Again, thanks for the selfless work you and your colleagues are doing. We wish you every success.

G.V.W. CA

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.

To whom it may concern:

Words cannot begin to express my appreciation for your wonderful articles and news. I am a 45 year old female. I could write a book on all my experiences over the years with CMT. But your recent articles have opened up a whole new world for me. Over the years I have taken so many of the medicines in your Medical Alert, which were prescribed to me by my many physicians. I had so many after-effects that they are too many to mention. I am so pleased that you are making all this information available to us. I would very much like to find one place to go that would treat everything.

I have gone to a rehabilitation hospital in NC which was a great help to me, but I feel I have learned more from your articles than from 35 years of trying to find information on my own. Maybe I'll be able to help direct someone else for help someday.

I have a 24 year old son that is just now developing a lot of the CMT symptoms. He is a professional artist and draftsman. I'm hoping that he will be spared some of the anxieties and frustrations in trying to find an informed physician thanks to your organization.

S.M. NC

Dear CMTA,

I am 25 years old and I was diagnosed as having CMT at age 4. I was lucky enough to have great parents and doctors who believed in fighting diseases. At age four I was put into leg braces and started on grueling therapy. The results were miraculous; my heel cords were stretched out and I became more and more athletic.After watching my recovery, the doctors became convinced that CMT could be controlled with VIGOROUS exercise. I started horseback riding in an English saddle to strengthen the muscles in my lower legs. At times it was extremely painful, but I was given the advice to keep my weight low and to exercise every chance I got.

At age 22 I was competing in Triathlons, sailing,horseback riding, playing tennis and golf and working out. There was no doctor who would believe that I had CMT. I was competing at levels above the normal person and had absolutely no signs of the disease. I strongly believe that the degeneration of peroneal muscles, caused by CMT, can be stopped. I hope there will be more studies and thought into the treatment of young CMT patients.

Unfortunately, I was in an automobile accident and had my legs and knees crushed, so I am not as asymptomatic as before. After numerous reconstructive surgeries, I am still on the road to recovery. After three years of trying to return to my prior level of activity, I am almost there. I am finally returning to running, biking,swimming,and sailing and will be competing in my first triathlon in the end of June. I have not returned to the competitive level I was at before the accident, but I am confident that with continued vigorous exercise I will.

I hope someone out there takes notice of how my doctors chose to attack the CMT problem so that other children may be as fortunate as I. After I graduated from college, I moved to the DC area and worked full time while putting myself through law school in the evenings. Given my schedule it was hard to fit in a lot of exercise, but the benefits far outweigh the sacrifice. Good luck CMT'ers, I hope I see some of you at my next road race or triathlon.

D.H. Washington,DC

(Editor's note: *This letter was edited for length only. The message was so encouraging that we wanted to print it, but were constrained by space limitations.)*

Recent Scientific Articles on CMT

"The Hand in Charcot-Marie-Tooth Disease"
Miller, MJ; Williams, LL; Slack, SL and Nappi, JF. **Journal of Hand Surgery** (British Volume, 1991) 16B:191-196

"Multi-Color Fluorescence, In Situ Hybridization and Pulsed Field Electrophoresis Dissect CMT1B Gene Regions"
Lebo, R.V. ; Lynch, E.; Wiegant, J.; Moore, K.; Trounstine, M,; van der Ploeg, M. **Human Genetics** (in press) 1991.

"Chromosome 1 Charcot-Marie-Tooth Disease (CMT 1B) Locus in FC Receptor Gene Region"
Lebo, R.V.; Chance, P.F.; Dyck, P.J. et al. **Human Genetics** (in press) 1991.

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 of 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 envelope indicating the geographic areas needed to: CMTA, 600 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.

¤

I want to be in touch!

Name: ______________________________
Address: ______________________________
____
Phone Number: ______________________________

Tell us about yourself:

☐ CMT Patient
☐ Interested Supporter
☐ Other
☐ Medical Professional
☐ CMT Family Member

Please check the appropriate boxes:

☐ Put me on the mailing list!
☐ CMT Neurology - $10
☐ Physical Therapy - $10
☐ Physical Therapy/Orthopedic Surgery - $15 (on one tape)
☐ Neurology/Genetics - $15 (on one tape)
☐ Send me a copy of the informative booklet "CMT Facts" - $3
☐ CMT Genetics - $10
☐ Orthopedic Surgery - $10

Join the fight against CMT!

Enclosed is my check for:

☐ $25 Member
☐ $________ other

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

Help Perpetuate The CMTA'S Work...

Remember the CMTA in Your Will

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

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

"I give and bequeath to the Charcot-Marie-Tooth Association, a not-for-profit corporation, organized under the laws of the Commonwealth of Pennsylvania, and having its principal office at Crozer Mills Enterprise Center, 600 Upland Avenue, Upland, PA 19015, the sum of $(____________) or (__________) percent of the rest, residue, and remainder of my estate to be used for general purposes of the Organization."

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

Great Lakes Area Registration Form

____
name(s) of attendees

____
address state zip

____
telephone

Fee for one person:	$21.00
Fee for two family members	$37.00
Fee for three family members	$48.00

Fees include lunch, coffee break, and afternoon refreshments.

Number of people attending: ______

Amount enclosed: $______

Return by Friday, September 6, 1991 to:

Patricia Dreibelbis
CMTA
600 Upland Ave.
Upland, PA 19015

You will receive confirmation of your reservation by mail.

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.

The following back issues of *The CMTA Report* are available at $2.50 a copy:

Summer '91
Winter '91
Fall '90
Summer '90
Spring '90
Winter '90
Fall '89
Summer '89
Spring '89
Winter '89
Spring/Summer '88
Summer/Fall '87
Spring '87
Winter '87

Write the CMTA (address below)

CMT...

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

..... is slowly progressive, causing deterioration of peripheral nerves which control sensory information and muscle function of foot/lower leg and hand/forearm muscles.

..... causes degeneration of muscles of foot, lower leg, hand and forearm.

..... causes foot drop walking gait, foot bone abnormalities (high arches and hammer toes), problems with hand function, balance problems, occasional lower leg and forearm muscle cramping, loss of some normal reflexes, occasional partial sight and/or hearing loss problems and in some patients may cause scoliosis (curvature of the spine).

..... may produce chronic pain and fatigue.

..... does not affect normal life expectancy.

..... has no effective treatment, although physical therapy and moderate physical activity are beneficial.

..... is sometimes treated surgically.

..... is usually inherited in an autosomal dominant pattern, which means if one parent has CMT there is a 50% chance of passing it on to each child.

..... is present in the world-wide population, with no apparent link to any one ethnic group.

..... may become worse if certain neurotoxic drugs are taken.

..... can vary greatly in its 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
600 Upland Avenue
Upland, PA 19015

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TO: