Charcot-Marie-Tooth Association

THE CMTA REPORT

This issue was partially funded by a grant from The Plastic Bottle Corporation

Summer 1990 **Vol.5/No.3**

THE GENETICS OF CMT

by Judith P. Willner, M.D. and Felice Yahr, M.S.

This article is taken from a letter to the mother of a newly diagnosed CMT child. At the time of the child's diagnosis appropriate testing was done on family members, and the child's mother, grandmother, great-grandmother and two maternal aunts were also diagnosed as CMT patients. Dr. Willner is a pediatrician in the Division of Medical Genetics at Mt. Sinai Medical Center, New York City and Mrs. Yahr, now retired, was a genetic counselor at Mt. Sinai Medical Center.

Charcot-Marie-Tooth disorder is a disease that affects the peripheral nerves. There are two forms of CMT which may appear quite similar clinically, since both may cause weakness of the leg and foot and foot deformities. In one form there is a loss of a substance called myelin. Myelin, which forms the sheaths surrounding the nerves, is necessary for normal nerve conduction. This rate of conduction is measured by an electromyogram (EMG). Thus with myelin loss, conduction velocity is slower. This lowered velocity, as revealed by an EMG, differentiates this form of CMT from the second form. This form is called Type I or hypertrophic CMT.

In the second form, while some nerves may be lost, there is no delay in the speed with which nerve impulses are conducted along the existing nerves. This form of CMT is called Type II or neuronal.

Both you and your mother show reduced conduction velocity. This permits diagnosis of the demyelinating form of CMT (Type I), and also tells us that in your family CMT is inherited in a dominant fashion. That is, inheritance of only one gene from one parent can cause the disorder to appear.

Felice Yahr

There is a recessively inherited form of the disease; in this case, an individual would have to inherit two genes, one from each parent, to have the disease. There is also an X-linked form of CMT where the CMT gene is located on the X chromosome. In that form the female carries the gene but does not have CMT. (However, some carriers may have very mild or sub-clinical signs of CMT.) That female carrier has a 50% chance of passing the CMT gene to a son and he will have the disorder. This female carrier also has a 50% chance of passing the CMT gene to a daughter and that daughter would be a carrier also. This CMT genetic pattern is similar to the inheritance of hemophilia.

By way of review, genes are the units of heredity. They are so small that they are invisible by any modern means of examination. Hundreds of genes together, always in a specific sequence, make up the structures known as chromosomes, which are present in every cell. With the exception of sperm and egg cells, all normal body cells have 23 pairs of chromosomes or a total of 46. When an egg and sperm come together at conception, they form a new individual who will have 46 chromosomes in all cells.

Every individual has many thousands of genes, each of which directs the cell to make a specific enzyme or other protein. Genes themselves are made of a substance which is capable of changing or mutating. Thus, among the vast number of genes present in all of us, a few in each individual are likely to have changed, with the result that they are unable to direct the production of (or code for) a normal protein. Each person is estimated to have three to eight such abnormal genes. The one which causes CMT probably should contain information for making a protein which is necessary for proper nerve function.

When a single abnormal or mutant gene in an individual causes the clinical picture of the disease as in your family, the disorder is said to be dominantly

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inherited. One parent carries a mutant gene for a dominant disorder and each child that individual has will have a 50% chance of inheriting the gene and therefore the disorder. It is true that sometimes a child will be born with such a disease when neither parent carries the mutant gene. In such a case the first mutation of that gene occurred spontaneously in the egg or sperm that form that individual. In such a situation the parents are not at increased risk to have another similarly affected child, but the affected child will a have a 50% chance to pass the mutant gene on to each of his/her children. We do not believe this the case in your family, since we have found evidence of the gene in you and your mother.

"The degree of severity of the disorder is impossible to predict."

Dominant disorders can vary widely in terms of the severity of the symptoms. This is called expressivity. In some individuals there can be severe clinical problems while other affected members of the family may have hardly noticeable or even unnoticeable symptoms. For example, in dominantly inherited deafness there may be a severely deaf child with a parent who has a very small hearing loss detectable only on a sophisticated audiogram. The parent may be unaware of this small loss. Therefore, in some of these dominantly inherited disorders while the gene is inherited, the severity of the symptoms may vary widely.

CMT is one of these. While the gene seems highly penetrant (that is an individual who carries it is likely to show some recognizable symptoms), the level of expressivity (degree of severity) is often low. In some families carrying this gene, the only visible sign of the disorder may be a high arch. In other families the severity varies and probably depends on whether other "modifier" genes were inherited along with the abnormal gene itself. The degree of severity of the disorder is therefore impossible to predict.

Since your daughter manifested symptoms earlier than you it is possible that she may not have received the modifiers that you have. When she herself has children each child will have a 50-50 chance of receiving the gene. Again the severity of the symptoms in a child with the gene will depend on the number of modifiers also inherited, some of which could come from the child's father.

For your information, it was mentioned that in the recessive form of CMT the parents of an affected individual appear to be completely normal and are normal by EMG. The normal gene that each parent has (paired with the abnormal recessive gene) protects them from having the disorder. This is the case with the pure recessive form of CMT inheritance. The inheritance of X-linked CMT we discussed earlier.

Although a biochemeical defect (that is, a lowered level of a particular enzyme or other protein) almost certainly exists in carriers of the recessive gene, no test has yet been devised to identify the carrier state. Recessively inherited CMT is generally a more severe disorder than the dominant form. It has an earlier age of onset and the symptoms progress much faster.

You were also told that while CMT is a progressive disease it usually progresses very slowly. Sometimes the condition appears so stable that it is misdiagnosed as polio. The very slow rate of progression of the symptoms in your mother is likely to be a familial characteristic of the disorder. Although this cannot be predicted with certainty, the same rate can reasonably be expected in your case as well as your daughter's. Your EMG showed that the rate of conduction of the sensory nerves was 35% below the normal mean and that of the motor nerves was 33% slowed. Latency (the time elapsed from conduction of the impulse to muscle contraction) was 50-60% below the normal mean. Yet your deficit is minimal and is unlikely to be apparent until you are much older. Like your mother, you may begin to experience some slight difficulty in later years.

The 50% chance in each future pregnancy for a child with CMT was discussed with you. It was also mentioned to you, as a matter of information, that every couple regardless of age or health, has a 2%, or 1 in 50, chance to have a child with a major congenital problem of some kind. Your 50% risk of a CMT child would be in addition to this general figure. We hope all of this is clear. Please contact us if you have any questions.

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Upcoming Conference

Mark your calendar!

New York, NY ..... Oct. 27,1990

Fame! Fame! Fame!

ATTENTION

CMT patients, families and friends...

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.

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
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.

Ask the Doctor

Dear Doctor,

I was diagnosed as having CMT approximately eight years ago. I am a 48 year old female. Other than muscle weakness I appear to be in good health with the exception of a bladder problem. The purpopse of this letter is to inquire if any other CMT patients have the same problem. The bladder appears to have had some nerve damage which results in my using intermittent catheters. This problem surfaced about five years ago. I know that this information is sketchy but my doctors are perplexed as to what caused the problem. I would appreciate any information that you may have regarding internal nerve damage caused by CMT or if this is even a possiblity. Thank you for your response.

G.B., IN

The writer is reporting inability to empty her bladder. She says that she is using intermittent catheterization and is presumable dry in between catheterizations. This type of difficulty is the result of inability of the bladder muscle to contract. The bladder is composed of smooth muscle like the bowel and is supplied by "autonomic" nerves that are not like the "somatic" nerves supplying skeletal muscle or the sphincter muscle controlling the base of the bladder.

The bladder muscle can be damaged by severe overfilling which stretches out the bladder muscle. This does not occur when the bladder has normal sensation because overfilling is painful. If there is loss of sensation in the pelvic area either because of postsurgical nerve injury or because of severe sensory neuropathy the bladder can overfill and be damaged without the painful sensation to signal the need to urinate. A neuropathy this severe may be seen in diabetics. Nerve damage may result from pelvic surgery.

I am not aware of patients with such serious sensory involvement as the result of Charcot-Marie-Tooth Disease. Occasionally medications can slow down bladder function. These are the benzodiazepines (Valium, Xanax), calcium channel blockers (used by cardiac patients), some muscle relaxants and tricyclic antidepressants such as Elavil or Tofranil.

Something to be considered is the coexistence of a spinal cord problem or possibility of a demyelinating disease. The patient's age would make these diagnoses unlikely.

Frances Dyro, M.D.

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Dear Doctor,

Recently I received a D & C (dilatation and curretage) in an outpatient day surgery center. There was quite a delay before surgery as the very concientious anesthesiologist felt that he was "coming in blind" regarding CMT and anesthesia. Unfortunately, his information center didn't have the information he needed to help with a decision about which anesthesia was best.

I didn't have any information except your list of drugs that are neurotoxic for CMT patients, and a warning about occasional decreased respiratory function. I had a spinal anesthesia, and everything went fine. However, I did have some minor siezure activity in the recovery room. They monitored my oxygen level with a sensor attached to my finger. It stayed at 91-93 range even though I received oxygen by nasal cannula. (I suspect that range is normal for me.) I was walking about 1 1/2 hours after the procedure. Subsequently, for two days after the procedure my legs ached terribly from my hips down to my ankles.

Was this due to the CMT, the spinal or the procedure?

Although many doctors are adamant that CMT does not effect respiratory function, I know it does. I am short of breath at the slightest exertion. Eight years ago, in biology class, (before my CMT diagnosis) my repiratory capacity tests were all lower than my classmates. I would like to know what type of anesthesia should I receive?

1. Can I have a general anesthesia?

2. Is the surgical area a determining factor?

Any information you can give me would be appreciated so that I can help the anesthesiologist the next time.

M.J., CA

Dr. James Garbern and Dr. Stephen Scherr of the University of Pennsylvania and Dr. Francis Dyro of the VA Medical Center, West Roxbury, MA were queried regarding the questions posed by this patient. We thank these noted neurologists for contributing their expertise.

Regarding the post operative pain, the doctors felt that the patient probably experienced a minor inflammatory reaction from the anesthesia. Dr. Dyro also suggested that the position the patient assumes for a surgical procedure can contribute to post operative discomfort. Continuing with the questions, CMT can affect respiratory function. Relevant to this and the questions posed about future surgery, each surgical procedure must be approached on an individual basis at the time of surgery. To a degree the surgical area does influence the choice of anesthesia, but again this decision would have to be made at the time of surgery. All physicians involved in any surgical procedure should be advised that the patient has CMT. The doctors furthered cautioned that drugs used prior to surgery for induction should not be a class of drugs called depolarizing agents, but rather should be curare drugs.

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Honoraria

In Honor Of...	By
Rebecca Sand	Lillian Raffel
Rebecca Sand	Sybil Whitman
Percy H. Sand	Minna Sand
Dr. Wm. J. Green	Susan Green
Ella Mae Mayers	Jeanell Haynie
Robert G. Bradwick	Sophia L. Bradwick
Kendrick T. McCann	Rebecca Sand
Sophie Bradwick	Faye Bradwick
R. Gordon Bradwick	Faye Bradwick
Joseph Wilcox	M/M William Usher
Rebecca Sand	M/M David J. Sarazan
M.D.A.	Francis & Lois Basquill

RESEARCH UPDATE

We checked in with Dr. Jeffrey Vance, a molecular geneticist at Duke University, Durham, N.C., about his on-going CMT research. Last year he and his colleagues had a major break through in that they identified chromosome 17 as having a CMT gene. Prior to this the known locations for a CMT gene were on chromosome 1 and the X chromosome. Dr. Vance, as well as researchers in many other locations, are continuing work on the chromosome 17 locus and are localizing the CMT gene to a smaller region of the chromosome. Dr. Vance believes that probably 75% of CMT cases will be attributed to the chromosome 17 gene.

Dr. James Lupski and Dr. Pragna Patel, both molecular geneticists at Baylor Medical Center, Houston Texas, are also working on the chromosome 17 gene with Dr. Carlos Garcia, a neurologist at Louisiana State University, New Orleans, LA. These researchers are studying several large French-Acadian kindred and are doing molecular studies and nerve conduction velocities (NVC). They are finding that the molecular approach to the problem (identifying the gene locus) and the clinical approach (NVC studies and symptoms the patient shows) need to be coordinated carefully in the research effort. In one large CMT kindred these researchers have tested family members who show no signs of CMT. However, some of these asymptomatic family members do indeed have abnormally slow NVC's putting them in the CMT diagnosis range.

We have spoken with many neurologists about the statistical incidence of CMT. All have agreed that at least 50% of CMT cases are undiagnosed. Drs. Lupski, Patel and Garcia's work seems to verify this opinion.

What does the work of the researcher mean to you the CMT patient? Ultimately, the locations of the CMT genes will be found. The long standing knowledge that CMT is not a single disorder but several differing disorders makes this task very difficult. Once the CMT gene(s) locations have been found genetic testing can be done. The protein(s) which the CMT gene(s) encode can be identified*. When the protein(s) are identified, then therapies and perhaps cures can be found. The opinion of the researchers is that this is still years away. However, the possibility exists that with luck a major break through will occur sooner. The history of scientific research does indeed contain significant discoveries that were made in conjunction with "Lady Luck".

***Editor's Note:** *See the lead article "The Genetics of CMT" for a good explanation of how genes, proteins and CMT intertwine.*

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For Your Information

Recent Journal Articles on CMT

The following references listed are recently published CMT articles in American and foreign scientific journals.

BENIGN AUTOSOMAL DOMINANT SYNDROME OF NEURONAL CHARCOT-MARIE-TOOTH DISEASE, PTOSIS, PARKINSONISM, AND DEMENTIA, by Tandan, R.; Adesina, A.; Sharma, K.; Fries, T.; and Pendlebury, W. Neurolgy, May 1990;40 (5):773-9

ISOLATION AND USE OF CHROMOSOME 1 PROBES FOR LINKAGE STUDIES ON CHARCOT-MARIE-TOOTH DISEASE, by Griffiths, L.R.; Zwi, M.B.; Mesterovic, N.; Ross, D.A.; Board, P.G.; Callen, D.F.; Mohandas, T.; Buckland, R.; Fletcher, J.M.; Driesel, A.J.; et. al. Annals of Human Genetics, January 1990;54 (Pt 1):31-7

GENETIC MAPPING OF AUTOSOMAL DOMINANT CHARCOT-MARIE-TOOTH DISEASE IN A LARGE FRENCH-ACADIAN KINDRED:IDENTIFICATION OF NEW LINKED MARKERS ON CHROMOSOME 17, by Patel, P.I.; Franco, B.; Garcia, C.; Slaugenhaupt, S.A.; Nakamura, Y.; Ledbetter, D.H.; Chakravarti, A. and Lupski, J.R. American Journal of Human Genetics, April 1990;46(4):801-9

HEREDITARY INDUCED PERIPHERAL NEUROPATHIES, by Lovelace, R., Clinics in Podiatric Medicine and Surgury, January 1990;7(1):37-50

NEUROPHYSIOLOGY OF FASTEST VOLUNTARY MUSCLE CONTRACTION IN HEREDITARY NEUROPATHY, by Logigian, E.L.; Hefter, H.H.; Reiners, K. and Freund, H.J. Archives of Neurology, January 1990;47(1):79-80

LINKAGE OF HEREDITARY MOTOR AND SENSORY SEUROPATHY TYPE 1 TO THE PERICENTROMERIC REGION OF CHROMOSOME 17, by Middleton-Price, H.R.; Harding, A.E.; Monterio, C.; Berciano, J.; and Malcolm, S. American Journal of Human Genetics, January 1990;46(1):92-4

PLASMA ALPHA/BETA LIPTPROTEIN COEFFICIAENT IN THE DIAGNOSIS OF PROGRESSIVE MUSCULAR ATROPHY, by Islamova, I.B., Lab. Delo, 1989;(10):34-5

X-LINKED RECESSIVELY INHERITED PERONEAL MUSCULAR ATROPHY, by Wang, D.S., Chung Hua Shen Ching Ching Shen Ko Tsa Chih, 1989 Jun;22(3):139-4 190

LOCALIZATION OF THE MUTATION IN AN EXTENDED FAMILY WITH CHARCOT-MARIE-TOOTH NEUROPATHY (HMSN I), by Raeymaekers, P.; Timmerman, V.; DeJonghe, P.; Swerts, L.; Gheuens, J.; Martin J.J.; Muylle, L.; DeWinter, G.; Vandenberghe, A.; and Van Broeckhoven, C., American Journal of Human Genetics, December 1989;45(6):953-8

ACUTE VINCRISTINE NEUROTOXICITY IN THE PRESENCE OF HEREDITARY MOTOR AND SENSORY NEUROPATHY TYPE 1, by McGuire, S.A.; Gospe, S.M.; and Dahl, G., Medical Pediatric Oncology, 1989;17(6):520-3

GENETIC LINKAGE OF THE AUTOSOMAL DOMINANT FORM OF CHARCOT-MARIE-TOOTH AMYOTROPHY AMD 3 GENETIC MARKERS ON CHROMOSOME 1, by Ferak, V.; Kadasi, L., Hrubisko, M.; Sivakova, D.; and Veghova, E., Cesk Neurol Neurochir, 1989 May;52(3):200-7

EPIDURAL ANESTHESIA FOR A PATIENT WITH CHARCOT-MARIE-TOOTH DISEASE, BRONCHIAL ASTHMA AND HYPTHYROIDISM, by Sugai, K. and Sugai, Y., Masui, 1989 May;38(5):688-91

CMT PATIENT PROFILE

The Signs Were There But Nobody Read Them

by Georgie Phalen

The following article is excerpted from a short story written by Georgie Phelan about her daughter Cathyann Marcoux. The perspective is that of a mother, and she eloquently tells her daughter's experiences. Cathyann's students are fortunate to have a teacher with such determination and the ability to adapt to what life brings.

Our daughter, Cathyann Marcoux, a 110 lb. dynamo and a source of constant moving energy, is what the world sees. Behind the facade her body is coping with the stressful disorder known as Charcot-Marie-Tooth disorder (CMT).

August, 1954 our beautiful baby daughter was born. Life went smoothly through the diaper, baby food and creeping stages. When she began to walk and run we noticed she had a balance problem causing her to fall often. Band aids and kisses brought her smile back and we attributed the problem to clumsiness.

At the age of five another symptom appeared. She had a heavy footfall. We were constantly telling her to pick her feet up when walking. She ran and played despite the tumbles she took from her ankles turning. Cathyann's pediatrician ordered sturdy oxfords and they improved her balance, but not her ego when she wore party dresses.

Life seemed normal and she entered jazz and modern dance classes. She enjoyed dancing immensely and signed up for ballet. The girls had scheduled training periods on the barre exercising legs and arms. A year went by and Cathyann earned her ballet slippers. Her eyes were sparkling with delight as she adjusted the sheepskin lining on her toes and tied satin ribbons around her ankles. What a thrill! I watched her face as she practiced in her prized slippers but her usual smile had been replaced. A taut expression and a face drained of color showed her pain. A real trouper, she came back repeatedly to practice, but finally admitted she couldn't function in the slippers because of extreme pain. (Years later after her disease had been diagnosed we realized a deformity, hammer toes, was already in progress. Pressure was on the bones of the foot instead of the fleshy pads of the toes.)

The author Georgie Phelan (L) and her daughter Cathyann Marcoux.

Cathyann had good years. She was able to ski, dance, work as a waitress and was quite athletic in high school and college. In college Cathyann pursued another of her dreams and received her BS degree in Elementary Education in 1976.

A fall wedding followed graduation and the ensuing year she began her teaching career. Her cup of life brimmed with love and happiness.

Cathyann was twenty-six when she became aware of serious pain in her feet. There was a loss of balance which caused her to stagger and fall. There were minor problems with hand function.

She was referred to a neurologist by her podiatrist and had a series of nerve conduction tests done. It was then she was diagnosed as having CMT.

Denial set in! She was young and full of life! How could she have a disease. An appointment was made at U/Conn Medical Center, Farmington, CT. The diagnosis was confirmed. Two young people clutched each other in fear. They had to learn to cope with the reality of this little known disease.

The first step was to establish herself where she would get good medical care. She had been a patient at Newington Childrens Hospital for treatment of scoliosis when in her teens. Cathyann opted to go there for treatment and follow-up progress because of the caring staff and their expertise. They are wonderful professional people who have guided her through leg brace adjustment, a muscle transfer-transplant, genetic counseling, therapy and dispensing of medication.

Cathyann devours all information regarding CMT and joined a local support group, as well as receiving and saving all issues of the CMTA Reports.

Her calf and thigh muscles are strong because of her dedication to exercising. The progression of this disease has affected her left hand and arm and she is losing balance control on the left side also. Doctors attribute it to the deformity of her left foot. Her foot rolls inward and she actually walks on the side of her foot causing incorrect body alignment and much pain.

An operation breaking the bones in her toes and foot and re-setting them correctly has been suggested. She would

(concluded on page 7)

Pepper Commission Reveals National Health Insurance Plan

The following articles are reprinted with permission from the Spring 1990 edition of Orphan Disease Update published by the National Organization for Rare Disorders (NORD).

On March 2, 1990 the Congressional Bipartisan Commission on Comprehensive Health Care (better known as the "Pepper Commission" after the late Congressman Claude Pepper), revealed its long awaited recommendations to Congress for a universal health insurance program and long-term (nursing home) care for all Americans.

The plan creates a two track system: health insurance through employers would remain in the private sector, and a "public plan" would replace Medicaid and be available to all citizens on a sliding scale fee basis. The goal would be to make health insurance more available, affordable and eventually compulsory.

Initially, the federal government would reform the private health insurance industry by enacting laws that would set minimum standards of coverage, prohibit denial of health insurance to people with preexisting health conditions, and prohibit insurance companies from charging higher premiums to people with a history of health problems. Tax credits and subsidies would be available to small businesses so they would be able to afford group health insurance policies. Self-employed people would be allowed to deduct 100% of their premiums from taxes.

As time proceeds, all businesses with more than 100 employees would be required to provide health insurance to workers and their dependents through private insurers or through the proposed federal public health insurance program.

For your records, our address is:

The CMTA
Crozer Mills Enterprise Center
600 Upland Avenue
Upland, PA 19015
(215) 499-7486

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

The Pepper Commission's "public plan" would provide coverage for employees and dependents not covered by other means, and non-working individuals who could "buy in" to the plan. If a person could not afford to pay for the premium, he or she would be subsidized. Physicians and hospitals would be paid according to Medicare rates. The program would be operated by the federal government even though states may opt to administer it.

The minimum benefits package would include primary and preventive care, physician and hospital benefits, and other services. People would pay a percentage of their medical costs (probably 20%) with a maximum limit on out-of-pocket expenses. The premiums of low income people (under 200% of the poverty level) would be subsidized.

The plan would be phased in over several years. Coverage for children and pregnant women would be made available immediately through the public plan. After five years, all Americans would be required to have health insurance.

Long Term Care

Severely disabled people of all ages would be eligible for this social insurance program that would provide home and community based care. The nursing home plan (NHP) would provide financial protection ensuring that no one would face impoverishment due to long-term illness.

People would be eligible to receive the first three months of nursing home care at no cost. This would allow those needing short nursing home stays to return home with resources intact. Both the federal and state governments would be responsible for shared financing of the NHP. However, states would administer the plan locally.

Although people would be encouraged to purchase supplemental private long-term care insurance to provide for any gaps in coverage, insurance companies would be regulated with standards set by the federal government. All benefits would be phased in over time.

Developing the Plan

The Pepper Commission plan for universal health insurance did not meet with bipartisan support. The Bush administration and many Congressional leaders are concerned about how much the plan would cost the government. It has been estimated that the program may cost $750 per person in the United States, which is substantially lower than the amount most people now pay for their present health insurance policy. However, the government must factor in subsidies for low income people and to date no analysis has been made of these costs.

Moreover, the "basic" health insurance plan, composed of minimum benefits, would not cover prescription drugs. It is assumed that many employers would enhance the minimum coverage by adding benefits that would not ordinarily be covered under the minimum plan. Thus details would have to be negotiated in Congress to determine just what "minimum" means.

Some Congressmen have suggested that the plan will never pass Congress due to its price tag and the federal deficit. Others believe, however, that the plan would not require extra funds because the government would simply redistribute the dollars that are presently being spent on our health care system. If enough people write to their Representatives and Senators showing interest in the Pepper Commission health plan, a dialogue may be initiated and a compromise worked out. Some in Congress feel that our nation is not yet ready for universal health insurance, but they don't have the catastrophic medical expenses associated with serious illnesses.

You may wish to write to your Representatives and Senators to stay informed on the latest developments and trends. For a copy of the Pepper Commission's recommendations contact: Senator John Rockefeller, Chairman, The Pepper Commission, United States Bipartisan Commission of Comprehensive Health Care, 140 Cannon House Office Bldg., Washington, DC 20515.

Editor's Note:

NORD and the CMTA are interested in learning of insurance problems encountered by patients with chronic and rare disorders. If you have had problems with insurance, please write to us here at the CMTA telling of your difficulties. If you have opinions about universal access to health care write to your representative and senators.

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Supreme Court Victory for Disabled Children

Reprinted with permission from NORD.

On February 20, 1990 the U.S. Supreme Court handed down a landmark decision in the case Sullivan vs. Zebley. NORD has been an active participant in this case, signing on to an Amicus brief on behalf of eleven-year-old Brian Zebley, a disabled child who had been denied Supplemental Security Income (SSI) and Medicaid by the Social Security Administration (SSA). Brian has congenital brain damage, mental retardation, eye problems and paralysis of his right side. In 1983 (Brian was age 4) the Social Security Administration had cut off his benefits saying he was not sufficiently disabled to qualify for SSI.

The Supreme Court made frequent reference to our Amicus brief filed on behalf of the American Medical Association (AMA), the American Academy of Pediatrics, the National Organization for Rare Disorders (NORD) and the Spina Bifida Association. The court ruled, in a seven to two vote, that disabled children are improperly being denied Social Security benefits by the Social Security Administration because child claimants are being held to a higher standard than adults. Children are eligible for SSI only if their disability matches or is "medically equal" to a small number of prevalent diseases on SSA's Listing of Impairments.

The high court determined that many rare disorders are not included on the crucial list, and children have been unfairly denied individualized functional assessments to determine their ability to perform age appropriate activities. Only about half of roughly 100,000 children who apply for SSI each year have been deemed eligible under current standards.

The brief submitted by the AMA, NORD and other organizations argued that SSA's Listing of Impairments is inherently incomplete and outdated. The great majority of rare disorders are not listed including such well-known disabilities as Down's Syndrome, AIDS, Muscular Dystrophy and Fetal Alcohol Syndrome. NORD supplied individual case histories (with names of the children deleted) to illustrate the gravity of this problem and the inequities of the system.

It is estimated that at least one-third of the children previously denied SSI and Medicaid will qualify for benefits as a result of this high court decision. In the future, children who apply for SSI will be given an individualized assessment to determine each child's functional capacity to perform age appropriate activities regardless on their diagnosis. All children who were denied SSI since 1983 because they were deemed not sufficiently disabled, should receive reconsideration of their claims.

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Addendum: The Social Security Administration (SSA) has convened experts in childhood disability to assist in the process of creating new regulations for SSI. If you wish to be on the mailing list for the SSI Childhood Disability Group contact: J. Kenneth McGill, Special Assistant, Social Security Administration, Baltimore, MD 21235 or call 301-965-3988.

Editor's Note

It is possible that some CMT children might now be eligible for aid under Social Security. Your local Social Security office can research this for you.

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ATTENTION CMT PATIENTS: Stand Up And Be Counted

Dr. James Lupski's CMT questionnaire appeared in the Spring'89 issue. If you have not filled this out, we urge you to do so now. If more copies for family members are needed, please send a stamped, self-addressed envelope along with your request to:

Karol Hitt/CMTA
Crozer Mills Enterprise Center
600 Upland Avenue
Upland, PA 19015

Completed forms may be mailed to the same address. Thank you.

Cathyann (cont'd from pg. 5)

be in a foot and leg cast for many weeks. She is concerned muscles will atrophy, and has been told CMT patients do not always make muscle comeback. Cathyann would appreciate any input regarding this operation and outcome. (Editor's note: If you would like to write to Cathyann about your experience with surgery, write to her care of the CMTA. We will forward your letter to her.)

Meanwhile, her motto is to live with hope rather than fear. No-one knows his/her future. Out daughter lives for the present, participates fully in life above and beyond as a teacher, teamleader, chaperone and friend to her students.

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THE CMTA GOES TO WASHINGTON

As an organization vitally concerned with a neurological disorder, the Charcot-Marie-Tooth Association was invited to present material at the National Meeting for Research in Neurological Disorders sponsored by the National Institute of Neurological Disorders and Stroke, a division of the National Institutes of Health. This meeting was conducted in Washington on April 17, 1990.

Ann Lee Beyer, a CMTA Board Member, presented a statement at this meeting. The material presented was prepared from the patient prospective and from the medical/research prospective. Information was provided by Dr. Robert Lovelace, Columbia University, Dr. Peter James Dyck, Mayo Clinic, Dr. Jeffrey Vance, Duke University and Dr. Roger Lebo, University of California at San Francisco. We thank these gentlemen for their cooperation and assistance.

During June the CMTA received from NIH the research directions for the 1990's, The Decade of the Brain. We are happy to report that CMT is mentioned specifically in the publication under the peripheral neuropathies.

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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.

San Diego, California
Contact: Gary Oleze (619) 792-1427

San Francisco, California
Contact: David Berger (415) 491-4801
After 6:00 pm

Connecticut
Contact: Linda Friedo (203) 374-8478

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

Parsons, Kansas
Where: Labette Community College
Parsons, KS
Contact: Tammy Taylor (316) 421-5268

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

Detroit, Michigan
Contact: Marrianne Tarpinian (313) 883-1123

Chicago, Illinois
Contact: Carol Wilcox (312) 445-2263

Cleveland, Ohio
Contact: Norma Markowitz (216) 247-8785

Boston, Massachusetts
Contact: Eunice Cohen (617) 894-9510

Central New Jersey
Where: Princeton Medical Center
Lambert House
Classrooms #1&2
Contact: Janet Selah (201) 281-6289

Northern New Jersey
Where: Englewood Hospital
Clinic Conference Room
350 Engle Street
Englewood, NJ 07631
Contact: Teresa Daino (201) 934-6241

Long Island, New York
Contact: Lauren Ugell (516) 433-5116

Westchester County, New York
Contact: Kay Flynn (914) 793-4710

Rochester, New York
Contact: Neale Bachmann (716) 554-6644
Bernice Roll (716) 584-3585

Delaware Valley, Pennsylvania
Meeting: Holy Redeemer Hospital
Meadowbrook, PA
Contact: Rex Morgan, Jr. (215) 672-4169

Pittsburgh, Pennsylvania
Contact: Garnett McDonald (412) 372-2853

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

Greater Atlanta, Georgia
Contact: Molly Howard (404) 253-5632

Western Georgia
Contact: Molly Howard (404) 253-5632

Orlando, Central Florida Area
Contact: Mary Beeler (407) 295-6215
Meeting: Third Saturday of every other month

Fort Pierce Area, Florida (Atlantic Coast)
Contact: Dorothy Stefanovich (407) 461-1016

VCR Tape Rental

The CMTA has available for rental four lectures which were taped at patient conferences sponsored by the Foundation. 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, Crozer Mills Enterprise Center, 600 Upland Ave., Upland, PA 19015.

October New York City CMT Conference

The CMTA is formulating plans for a CMT patient/family conference at Columbia University Medical Center on October 27, 1990. The meeting will be in the Zabriskie Auditorium which is on the first floor of the Neurological Institute, 710 West 168th Street, New York City.

The program will start at 9:30 AM with registration. Dr. Robert Lovelace will begin the program with an introductory explanation of CMT and then answer questions from the audience. Dr. Lovelace, a professor of neurology at Columbia College of Physicians and Surgeons and co-director of the neuromuscular clinic, is an expert on Charcot-Marie-Tooth disorders. Following Dr. Lovelace will be Matthew Gibble, a physical therapist. Mr. Gibble, a private practitioner, is actively involved in the treatment of CMT patients.

Following a box lunch the afternoon program begins with a talk by Dr. Gerald Webber, a podiatrist. Dr. Webber is affiliated with the New York College of Podiatric Medicine. The rest of the program will consist of a psychologist, geneticist and a physiatrist. The psychologist will address the issue "Adapting to CMT." Each of the speakers will allocate time in his/her presentation for questions. This is a wonderful opportunity to ask the experts.

There is no fee for the meeting, but there is a fee for the box lunch. Therefore, if you plan to attend please complete and mail the conference reservation form. You are welcome to come without making a reservation, but we will be unable to supply you with a box lunch. Eating facilities are scarce in this area. Parking facilities, for a fee, are available and the building is handicap accessible.

The cost of the box lunch is $6.00 and reservations, including a check for lunch if desired, must reach us by **October 24, 1990.** If you have questions please contact Ann Beyer, 18 Brownstone Way, Upper Saddle River, NJ 07458 or call 201-391-4624.

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New York Conference Reservation Form

NAME_____

STREET_____

CITY_____

STATE & ZIP_____

☐ YES, I will attend and want ___ box lunches at $6 each. My check for $_____ is enclosed.

☐ NO, I do not want a box lunch but I will attend.

Must reach us by October 24, 1990

Mail To: ANN LEE BEYER
18 Brownstone Way
Upper Saddle River, NJ 07458

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, Crozer Mills Enterprise Center, 600 Upland Ave., Upland, PA 19015.

Honorary Gift

In honor of: (person(s) you wish to honor)

_____

Send acknowledge to:

Name: _____

Address: _____

_____

Occasion:

☐ Birthday
☐ Wedding
☐ Anniversary
☐ Holiday
☐ Thank You
☐ Other

Memorial Gift

In memory of: (name of deceased)

_____

Send acknowledge to:

Name: _____

Address: _____

_____

Amount Enclosed: $_____

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

Gift Given By:

Name: _____

Address: _____

_____

The CMTA Goes To NORD

Karol Hitt (L) and Ann Lee Beyer (R) with Abbey Meyer, Executive Director of NORD at the May Conference.

Washington, D.C. was the site of the National Organization of Rare Disorders (NORD) annual meeting. The CMTA is a full voting member of NORD and was represented by Karol Hitt, CMTA President, and Ann Lee Beyer, a CMTA Director. The meeting is an opportunity to learn what is taking place on the national level regarding health care, research funding, and orphan drug development and production. Additionally, NORD's program of workshops and informational panels are a great way to learn and to network with other voluntary health organizations. The workshops are true working situations, and the information learned and contacts made are invaluable.

Abbey Meyers, executive director and a founder of NORD, is a tireless worker for the patient with a rare disorder. This organization has been very actively involved in educating members of Congress as to the plight and needs of persons with a rare disorder. Included in NORD's current area of interest is the state of health insurance in the United States, particularly as it affects the person with a chronic/rare disorder. NORD is to be commended for a job well done.

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LETTERS

We want to hear from YOU!

Write us at:

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

The CMTA Report is published by the Charcot-Marie-Tooth Association, a tax exempt not-for-profit corporation.

Published & distributed by:
Educational Marketing Services
P.O. Box 760
Hoarce Harding Station
New York, NY 11362
Tel. 718/279-8516

Layout by:
Chesapeake Bay Design
48 Henry Court
Hollywood, MD 20636
Tel. 301/373-5912

MONEY MONEY MONEY

We all need it and the CMTA is always looking for ways to fund our program. We have found two wonderful sources, but we need your help.

The first is the **United Way.** Through the Donor Choice Program you can designate the Charcot-Marie-Tooth Association (formerly the National Foundation for Peroneal Muscular Atrophy) as the recipient of your pledge. When you make your pledge at work this fall write in our name. We are eligible for these funds because we are an IRS tax exempt non-profit organization. Spread the word to friends and family and ask them to also designate the CMTA as their Donor Choice.

The second source is the **Combined Federal Campaign (CFC).** We recently were approved by the CFC and now are on its list. This means that any employee on the federal payroll (military or civilian) can now designate the CMTA as the recipient of his/her pledge. **Our CFC number is #1031.** We are very pleased with this designation as the federal government has very exacting standards for inclusion in this list. Again, spread the word to friends and family who are a part of the federal employee system. Ask them to designate the CMTA.

We also applied for and received CFC approval from the Philadelphia area Local Federal Coordinating Committee. Not only will we be in the book for the National CFC campaign, but also listed on the local level during the fall 1990 campaign in the Philly area.

Memorials

In Memory Of...	By
Mildred Schomp	Barbara Jeager
Gertrude Colgan	Union Carbide Corp.
Dr. John Valenti	Mrs. Harold C. Lewis
R. Sullivan	Edward Scarsella
Vivian Ramsey	Myrtle Brimhall
I.. S. Warner	Bob Warner
Andrew Lane	Clarence Vater

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 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, Crozer Mills Enterprise Center, 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.

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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
- ☐ CMT Genetics - $10
- ☐ Orthopedic Surgery - $10
- ☐ Physical Therapy/Orthopedic Surgery - $15 (on one tape)
- ☐ Neurology/Genetics - $15 (on one tape)

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.

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For the CMTA

This material is presented for educational purposes only and is not meant to either diagnose or prescribe. While there is no substitute for professional medical care for Charcot-Marie-Tooth Disease, these briefs offer current medical opinion that the reader may use to aid and supplement a doctor's treatment.

Attention CMT Patients!

Dr. James Lupski, of Baylor Medical Center, requests that CMT patients who have a second inherited condition contact him. Please, when you write give the name of the second condition. Also, CMT patients who have a known chromosomal anomaly are asked to contact Dr. Lupski at the CMTA, Crozer Mills Enterprise Center, 600 Upland Avenue, Upland, PA 19015. (215) 499-7486.

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:

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

Write or call the CMTA
215-499-7486

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 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.

THE CMTA REPORT

information on Charcot-Marie-Tooth disease from the

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

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