National Foundation for Peroneal Muscular Atrophy

THE NFPMA REPORT

Winter 1987 **Vol.1/No. 1**

Providing information on Charcot-Marie-Tooth disease (or Peroneal Muscular Atrophy), the most common inherited neurological disease

ABOUT THIS FIRST ISSUE:

The National Foundation for Peroneal Muscular Atrophy publishes this, our first report, with several goals in mind.

Research and the ultimate treatment of Peroneal Muscular Atrophy (also known as Charcot-Marie-Tooth disease, or CMT)are primary goals of the Foundation. Founded in 1983, the NFPMA hopes to insure continued, long-term support for research on this disorder. Recent developments in gene mapping studies, preliminary experimental drug trials and in the findings of urinary organic acid metabolic screening have given us cause for optimism. This report will enable us to keep our readers informed of current research and to insure that recent developments will readily be applied to the investigation and treatment of CMT.

The NFPMA is also responding to the need of CMT patients, families, and the medical community for better information. There are few resources available to gain the knowledge and understanding necessary to deal effectively with this disease. CMT is too often misdiagnosed, frequently misunderstood, and patients repeatedly misdirected.

This report will be sent to the medical community, thus enabling them to better serve the needs of the CMT patient. We will also share practical suggestions and ways to better cope with the disabilities connected with CMT.

Finally, the NFPMA eventually hopes to establish a system of patient support groups throughout the country.

The goals of the NFPMA are high. Research is the only hope for a cure and we are closer than ever before, in part because of this organization's tenaciousness and your generous support.

Please send us your comments and suggestions.

PHYSICAL THERAPY: How To Find It and Pay For It

The primary functional effect of Charcot-Marie-Tooth disease (CMT) or Peroneal Muscular Atrophy (PMA), is the loss of muscle strength, particularly below the knee and in the hands. This loss of strength makes walking more difficult and tiring. Weakness in the hands can often result in difficulty in writing and in performing tasks around the house. In addition to the direct effects of weakness, unaffected muscles tend to become abnormally tight because they no longer are opposed by balancing muscles. This tightness may result in deformities in the feet and hands, making activities even more difficult and often painful.

While there are no cures for this disease, there are ways to slow the progress of the weakness and the resulting deformities. Appropriate exercises may be used to strengthen the weakening muscles and to stretch those muscles that are becoming tight.

Dr. Carol Oatis (right) counsels a CMT patient

IT IS VERY IMPORTANT THAT EACH MUSCLE IS EXERCISED CORRECTLY. This exercise program should be developed under the supervision of a knowledgeable physical therapist.

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Publication of this edition of the NFPMA Newsletter has been partially supported by P. W. Minor & Son, Inc., makers of regular and orthopedic shoes since 1867.

NFPMA SPREADS MESSAGE AT REGIONAL CONFERENCE

In June 1986, the National Foundation for Peroneal Muscular Atrophy hosted its second Philadelphia regional conference on Charcot-Marie-Tooth disease for patient family members and medical professionals at Wyndham Alumnae House, Bryn Mawr College. This conference was one of several that have been held within the last two years to inform and organize CMT patients in the Philadelphia, New York, and Washington, D.C. areas. Speakers at these conferences have been health care professionals who treat and/or study CMT disease as well as some members of the patient community with information to share. Previous NFPMA educational symposia have been held at the Bethesda Holiday Inn (Bethesda, MD), the International House (Philadelphia), Mount Sinai School of Medicine (New York), and the Neurological Institute of Columbia University (New York).

The Bryn Mawr meeting, like the previous conferences, gave patients and their families an opportunity to personally meet with medical professionals who are familiar with their disease and to participate in a unique educational forum focusing entirely on their CMT problems. The open exchange of information between the health care community and its public and the patient-to-patient contact available at the conferences have become an important part of the NFPMA program.

Generally the topics covered at a regional conference will include orthopedics, neurology, genetics, physical therapy, and current CMT research. Such was the case at Bryn Mawr, but if a theme dominated the lectures that day it was certainly the message of the importance of physical therapy to the CMT patient.

Richard Bowen, M.D. of the Alfred I. Du Pont Institute of Children's Hospital in Wilmington, Delaware spoke of orthopedic options for the CMT patient. Dr. Bowen is a pediatric orthopedist who has worked with a large population of children with CMT disease. His approach to orthopedics and the CMT patient was an enlightened and multi-disciplinary one. Remarking that most CMT people do not need a lot of surgery, Dr. Bowen stressed that ortbopedic surgery cannot be a cure, nor stop the process of CMT (a breakdown of nerves), but it may alter some resultant bone and muscle damage and help return some function to a patient. Dr. Bowen has been successful with the triple arthrodesis plus tendon transfer, which is a surgical technique that can help to keep the CMT foot level and restore ankle flexing or the "up and down" movement needed for walking.

In describing the triple arthrodesis and tendon transfer Dr. Bowen emphasized the importance of the patient being in good physical condition before the procedure and pushing to keep in shape during the long recovery period. Although the procedure involves a lengthy healing time (3-6 months), the CMT patient must keep moving and avoid the urge to recuperate in bed. The more someone with CMT lies down to recover, i.e. if they indulge themselves in post-operative "over-pampering," the more strength they will lose. Therefore the motto "Use it or lose it" is fundamental for the CMT patient.

In a disease such as CMT where there is no current cure, physical therapy ranks as the closest thing to consistent treatment. Carol Oatis, P.T., Ph.D., a partner in the Philadelphia Institute for Physical Therapy, spoke at the June conference about the benefits of physical therapy (PT) for the CMT patient. She outlined the goals of the patient in PT as an effort to maintain strength, diminish the risk of falling, conserve energy, and minimize deformity problems. According to Dr. Oatis, physical therapy should be offered early since it often buys time against the progression of CMT. A good exercise program can be set up by a therapist to address the specific weakness of the CMT patient and improve overall condition.

Following Dr. Oatis at the podium was Dr. Erlinda Reyes, a neurologist at the Temple University School of Medicine, who has since relocated to New York. Dr. Reyes described the four main features of CMT (inherited, neurogenic, localized in lower extremities, slowly progressive) and provided a thorough treatment of its two main presentations- muscular atrophy and deformities of the foot and hand. Dr. Reyes also spoke of the importance of establishing a linkage between the disease and a genetic marker to allow very early detection or even pre-symptomatic diagnosis. Her remarks and those of the other speakers underscored the problem of misdiagnosis of CMT, causing patients to wander among doctors who are unfamiliar with this most common inherited neurological disease.

CMT patient Joan Horvath echoed Dr. Reyes' concern over misdiagnosis claiming, "Misdiagnosis follows this disease around like a shadow." Ms. Horvath then spoke convincingly about her personal experience with physical therapy and the importance of being an assertive patient. She is a walking, tennis-playing testimonial to the benefits of physical therapy. One would be hard-pressed to notice that Joan is afflicted with a physical disorder since, through her PT efforts, she has been able to retain an amazing amount of function.

At one time approaching a toll booth could make Joan break into a cold sweat fearing the difficulty she would have picking up the necessary coins, or perhaps not feeling the change returned to her

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Peroneal Muscular Atrophy (CMT) . . .

. . . is the most common inherited neurological disease, affecting approximately 125,000 Americans.
. . . is also known by its historical name, Charcot-Marie-Tooth Disease, for the three doctors who first reported on it in 1886.
. . . is slowly progressive, causing deterioration of peripheral nerves which control sensory information and muscle function of lower legs and forearm voluntary muscles.
. . . causes degeneration of peroneal muscles (located on the front of the leg below the knee) and subsequent atrophy of additional lower leg and forearm muscle groups.
. . . 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 in more severe cases may cause scoliosis.
. . . does not affect normal life expectancy.
. . . has no effective treatment, although physical therapy and moderate physical activity are beneficial.
. . . is usually inherited in an autosomal dominant pattern, affecting half the children in a family with one PMA parent.
. . . is present in the world-wide population, with no apparent link to any one ethnic group.

PHYSICAL THERAPY

(continued from page 1)

Physical therapy is a branch of the health care system which specializes in movement disorders resulting from such diseases as CMT. Physical therapists are specifically educated in the evaluation and treatment of muscles, nerves, and motion. Like most health professionals, physical therapists often specialize, so the following is a guide to finding a physical therapist (P.T.) who can help you develop the right program of exercise.

What qualities should you seek in a physical therapist? Most physical therapists will have at least heard of CMT. Those who work with patients who have muscular dystrophy will probably be the most familiar with the disease. However, there are more important criteria in choosing a physical therapist than his or her experience with CMT. Because the disease most often manifests itself in weakness of the legs and feet, it is most important to find a therapist who will do a complete evaluation of the muscles of the legs and feet and who understands the mechanics of the feet and legs during locomotion. Such an evaluation should take approximately one hour. The physical therapist should then be able to develop a program of exercises which you can perform at home or a more intensive program in a physical therapy department. In addition, the therapist may find that you need braces or splints to support your hands or feet in order to keep them in proper alignment.

The physical therapist should also be able to evaluate the muscles of the hands. If there is significant involvement of the hands, it may be necessary for the therapist to refer you to a hand specialist who may be either a physical or occupational therapist.

Finally your physical therapist must be someone you can trust and with whom you can work. Physical therapy requires consistent and hard work. In order to be willing to put out such effort, you must be comfortable with the therapist who should serve as your teacher, coach, and cheerleader.

Now the question is where to go to find such a physical therapist. One of the best sources is a "satisfied customer." Talk to other patients with CMT who have seen a physical therapist. If they are satisfied, talk to their therapist. Even if the therapist is not located nearby, he/she may know of a therapist in your community.

Most hospitals have a physical therapy department (rehabilitation medicine) and many physical therapists practice privately. The telephone directory should provide information about all of these facilities. Once you have found a facility close to you, talk to the chief physical therapist. Ask if anyone there has experience with CMT and find out if anyone has a special interest in locomotion and/or feet. What does an initial visit entail and how long will it take? If you still have not found adequate assistance, contact the local chapter of the Muscular Dystrophy Association, the American Physical Therapy Association (1111 N. Fairfax St., Alexandria, VA 22314; phone: 703-684-2782), or the department of rehabilitation medicine at a regional medical school.

Once you have found a physical therapist, the expense questions remain. How much will it cost and who will pay for it? Evaluations can cost between $30 and $100. Return visits may range from $20 to $75. The price range is very broad and not necessarily a good indicator of quality.

Most insurance policies cover at least some portion of the cost of physical therapy. Blue Cross Major Medical usually covers 80% of the remaining cost following payment of the deductible. Private insurance carriers may cover more or less of the cost. Medicare will pay for progressing physical therapy services but not for maintenance or long-term care for chronic illnesses. Medicare also pays different percentages of the bill depending on the setting in which you receive services. Medicaid provides different services in each state. Your therapist should be able to help determine the extent of your insurance coverage. The Muscular Dystrophy Association will provide some assistance for physical

(Continued on page 9)

METABOLIC SCREENING STUDY POINTS THE WAY TO A BETTER UNDERSTANDING OF CHARCOT-MARIE-TOOTH DISEASE

A variety of problems confront the medical scientist interested in Charcot-Marie-Tooth disease. To begin with, even patients in the same family may show somewhat different degrees of medical symptoms. Patients from different families may have diseases which appear similar but actually have different underlying biochemical causes. Examination of patient nerve and muscle samples under the microscope reveals disease-related changes which are not unique just for CMT and which do not readily suggest what the basic health problem(s) may be. Of the relatively small amount of biochemical information available on CMT patients, most such measurements give values similar to those obtained from healthy people. This indicates that scientists have not yet identified which areas of cellular metabolism are most critically affected by CMT. In addition, while pieces of peripheral nerve may be obtained for brief experiments, this kind of cell cannot be kept alive and grown in the laboratory, as can be done with many other types of cells.

Of course, from the CMT patient's point of view there are other problems. Donating a muscle or nerve biopsy sample is actually minor surgery. No one looks forward to invasive procedures of this kind.

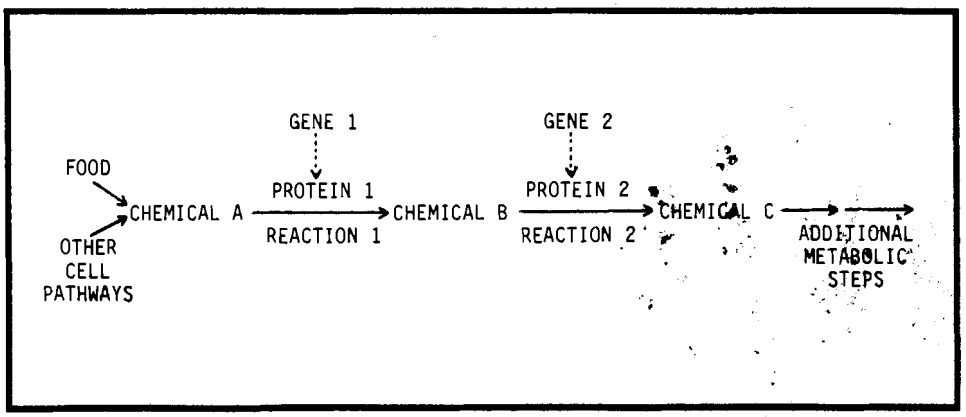
During June of this past year my colleagues and I completed a biochemical research study on a CMT family which was designed to get around many of the problems normally associated with research on this disease. Working in laboratories at the University of Pennsylvania Medical School, we have applied a type of research technology known as gas chromatography/mass spectrometry (GC/MS) to the study of CMT patients. As this study was based on analysis of urine samples, patient participation required no special medical procedures. GC/MS is the most sensitive and comprehensive tool now available for physiological studies. From a sample extract corresponding to one drop of urine we can measure the amounts of approximately 150 metabolites, giving us a broad overview of each patient's physiological status. Our recently completed study appears to be the most thorough application of this technology to any of the inherited neuromuscular diseases, involving analysis of samples from six CMT patients from the same family and analysis of corresponding data obtained from six normal volunteers.

Of the 150 metabolites analyzed in our GC/MS study, known as organic acids, only three showed characteristically altered concentration levels in our CMT patients. Our patients showed average levels of each of these metabolites corresponding to only 1/3 t o 1/2 the levels seen in normal subjects. As these three organic acids have quite similar chemical structures, our findings may indicate that their decreased concentration levels reflect a single common metabolic problem. These characteristic metabolites are known as furancarboxylic acids. In the body they are derived from precursors known as furanaldehydes, which are present in many foods. Decreased excretion of furancarboxylic acids in CMT patients may indicate that furanaldehydes are not being disposed of properly and may accumulate in the body. Furanaldehydes have chemical structures similar to those of organic solvents known to cause damage to peripheral nerves. Further work is needed to confirm and extend the results that we have obtained so far. Nevertheless, it is not too soon to describe this unusual research effort to the CMT patient community and to discuss the the opportunities that it offers.

THE BIOCHEMICAL BASIS OF LIVING ORGANISMS

The human body consists of millions of living subunits known as cells. Cells may be seen with the aid of a microscope. Each has its own enclosing membrane and two complete sets of human genes (except for certain reproductive cells). The cells of different body organs are unique, each type reflecting the expression of a distinct sub-set of genes. Hence not all genes are expressed in each cell type, only those needed for each organ's specific role.

But just how do genes control cell functions? The genetic message contained in the unique code of each gene determines the structure of a particular protein. In general, each protein serves as a catalyst which facilitates one single step in cellular metabolism. Overall, a living cell contains several thousand proteins, each having its



own unique chemical task. However, these various chemical transformations are not a random collection of reactions. Instead the various protein-mediated chemical reactions are organized into specific biochemical pathways. These organized sequences of chemical reactions enable each cell to digest food and transform food components into whatever unique biomolecules are needed to continue life, including new copies of genes and proteins. A generalized example of a metabolic pathway is shown on page 4.

Each single chemical transformation is catalyzed by a specific protein. The product of one chemical step is the starting point for the next. Typically all living cells, whether they be animal, plant, or microorganism, share more or less the same "core" biochemical pathways which perform routine life functions such as processing food molecules, with each cell type also expressing additional specialized biochemical pathways.

THE METABOLIC BASIS OF INHERITED DISEASE

Volumes have been written on this topic, but perhaps the most fundamental concept can be illustrated by referring to the general metabolic pathway noted on the previous page. What if there were a physical defect in the gene that contained the code for protein two? Perhaps an error has occurred in the sequence of the genetic code. Perhaps a piece of the gene is actually missing. The result would be a structurally altered protein two lacking biological activity, or simply a decreased amount of the protein. In either case a kind of bottleneck would develop on this biochemical pathway at reaction two. With little or no functional protein two available, its normal catalytic role will remain unfulfilled. Protein one will continue to form chemical B, leading to the accumulation of chemical B and possibly that of its precursors also. In this state the metabolic pathway is effectively blocked and chemical intermediates which are normally present in only trace amounts now accumulate. Many such chemical intermediates are toxic at high concentrations, causing adverse effects on a variety of cellular metabolic functions and leading to one or another state of disease.

METABOLIC SCREENING BY GC/MS

Trace amounts of metabolites leak out of cells and into the blood. In the kidneys these metabolites pass from blood to urine. Thus if we had an appropriate sophisticated laboratory procedure we could measure trace amounts of numerous cell metabolites from a single urine sample. The result would be an overview of a person's health at the physiological level, a detailed form of metabolic screening.

Only within the past ten years has such laboratory methodology become available. The procedure known as gas chromatography/ mass spectrometry is capable of separating more than 100 cell metabolites generally referred to as organic acids, then identifying and measuring the amounts of each. Organic acids are relatively small size biomolecules, much smaller than proteins or genes. Each has a common characteristic functional group as part of its structure known as a carboxylic acid group, consisting of one carbon, two oxygen, and one hydrogen atom linked together.

There are four basic steps involved in metabolic screening by GC/MS: preparation of the sample; separation of organic acids by gas chromatography; recording the data on each organic acid by mass spectrometry; and final data analysis. In the first step the organic acids are extracted from the original sample by mixing with an organic solvent, followed by removal of the solvent by evaporation. The resulting extract is then chemically derivatized to yield modified organic acids which are more volatile and thus easier to analyze.

Each sample is then injected into a gas chromatograph/mass spectrometer. Based on molecular size and volatility, organic acids emerge one by one from the gas chromatograph and enter the mass spectrometer. Here the molecules of each organic acid are shattered with a high energy electron beam, giving a collection of fragments unique for each metabolite. The number and size of these fragments are then recorded on a computer disc. For each metabolite, measuring the amount of the most commonly seen ion fragment provides an estimate of the metabolite's concentration.

PUTTING THINGS IN PERSPECTIVE: What CMT patients may gain from research of this kind

GC/MS technology is the most comprehensive and efficient tool currently available for defining the health of CMT patients at the physiological level. In one experimental procedure approximately 150 metabolites can be measured from each patient. As this procedure is based on analysis of urine samples, invasive medical procedures such as nerve or muscle biopsies are unnecessary.

No single research study answers all questions about a disease. Yet the results of our first GC/MS project suggest that we now have an exciting opportunity to gain a detailed understanding of the cause of this disease. In the family we have examined, CMT is inherited as an autosomal dominant trait. Since people have two genes for any one genetic trait (one from each parent), these patients have one CMT gene and one corresponding normal gene. This suggests that whatever the physiological basis of this disease may be, only a partial metabolic block is present, which should result in a loss of approximately half of this metabolic function. The findings reported in our first GC/MS study suggest that CMT patients in this

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STUDY *(continued from page 5)*

family may have a partial metabolic block at the level of protein mediated oxidation of furanaldehydes to corresponding furancarboxylic acids. If so, furanaldehydes may accumulate in the body.

Confirmation of these preliminary findings would provide the first opportunity to partially limit the crippling effects of this disease by defining a diet low in furanaldehydes. For the first time research scientists would be able to investigate possible drug therapies based on a knowledge of what causes CMT. In coming months the National Foundation for Peroneal Muscular Atrophy will continue to work with scientists at the University of Pennsylvania Medical School to confirm and extend the results of our first GC/MS metabolic screening study.

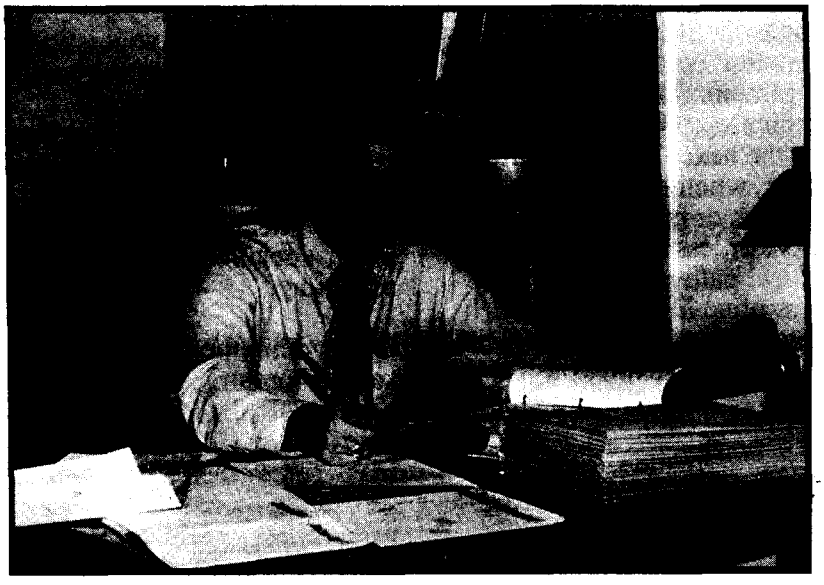
Our special thanks to Dr. Howard Goldfine of the University of Pennsylvania Department of Microbiology and to Dr. G. Clare Kahn of the University of Pennsylvania Department of Pathology and Laboratory Medicine for providing facilities used in our GC/MS studies and advice. The work described in this report was supported by the National Foundation for Peroneal Muscular Atrophy and by a Public Health Service fellowship to Dr. Shapiro.

Preliminary urinalysis work from 1981 to 1983 was supported by a fellowship to Dr. Shapiro from the Muscular Dystrophy Association, by an earlier Public Health Service fellowship to Dr.Shapiro, by the University of Pennsylvania Medical School Biomedical Research Support Grant, by grants from the Muscular Dystrophy Association and the American Academy for Cerebral Palsy and Developmental Medicine and by gifts from Frank T. Crohn, Sr. and Nancy and Howard Finkelman to the NFPMA.

The technical assistance of Lois Murphy is gratefully acknowledged, as is the cooperation of Drs. Fred Karush, Donald Young, and Leslie Shaw.

ABOUT THE AUTHOR

Howard Shapiro, Ph.D., is a CMT patient who has devoted much of his career as a research scientist to the study of Charcot-Marie-Tooth disease. He is one of the founders of the NFPMA and serves as its secretary and director of scientific program.



Dr. Howard K. Shapiro working on calculations for metabolic screening study

Dr. Shapiro received his B.A. in biology from Franklin & Marshall College in Lancaster, PA (1970) and his Ph.D. in biochemistry from Bryn Mawr College (1977). His professional experience includes work as a research chemist in 1977-78 with Dr. Joseph Rabinowitz at the Radioisotope Laboratory of Veterans Hospital in Philadelphia. Post-doctoral research fellowships from the Muscular Dystrophy Association and the National Institute of Health allowed Dr.Shapiro to work during the period of 1978 to 1985 with Dr. Robert Barchi, Dr. Howard Goldfine, and Dr. Fred Karush at the University of Pennsylvania.

Most importantly, as you can read in this issue, Dr. Shapiro has devoted much of his energy since 1981 to the application of gas chromatography/mass spectrometry in the study of CMT in an effort to solve the physiological puzzle of CMT disease.

When he is not immersed in CMT research, Dr. Shapiro keeps busy with a relentless schedule of trips, conferences, appointments, grant applications, fund-raising, and plans on behalf of the National Foundation for Peroneal Muscular Atrophy.

CORPORATE SUPPORT HELPS NFPMA CAUSE

The National Foundation for Peroneal Muscular Atrophy wishes to gratefully acknowledge a recent $10,000 grant from the Revlon Corporate Foundation of New York. This grant is one of many made by the Revlon Corporate Foundation in recent years to support womens' issues and help fight birth defects.

This generous contribution will serve to partially support the continuation of Dr. Shapiro's CMT research studies at the University of Pennsylvania Medical School.

The Not-Your-Neighborhood Shoe Store

Every CMT patient has his or her own foot story. For some patients buying shoes is only a minor problem, but for others it can be quite a dilemma. Some patients have such problems getting comfortable shoes that they'll keep each good pair for years, coming to measure time in their lives in "shoe eras." Bad shoes can make every step painful, usually because the shoes are not wide enough. Listed below are several sources for special shoes. Here again please let us know if you have additional shoe ideas.

Manufacturers—These companies will answer calls and refer the caller to the nearest store carrying their shoes.

P.W. Minor & Son,Inc.
3 Treadeasy Avenue
P.O. Box 678
Batavia, NY 14020
(716) 343-1500
(Full line of orthopedic shoes)

Sabel Shoe Company
Benson East Building
P.O. Box 644
Jenkintown, PA 19046
(215) 885-1772
(Full line of orthopedic shoes for men, women, and children)

Musbeck Shoe Company
Oconomowoc, Wisconsin 53066
(414) 567-4416
(Full line of orthopedic shoes for men and women)

Orthopedic Shoe Repair
Nelson, Inc.
1221 6th Avenue
New York , NY 10020
(212) 737-3984

TSR Shoe Corporation
Nelson Shoe Repair
1221 Avenue of the Americas
New York, NY 10020
(212) 869-3552

The following are retail stores:
Rochelle Molded Shoe Company
712 Foulk Road
Wilmington, DE 19803
(302) 762-2939
(Custom made shoes for men, women, and children)

Davis Shoe Therapeutics
3921 Judah Street
San Francisco, CA 94121
(415) 661-8705
(Custom and ready-made shoes for M and W; by appointment only; some children's styles; new line—brochure is available)

Louis Carlascio
283 Grove Street
Jersey City, NJ 07302
(201) 333-8716
(Custom and ready-made shoes for M and W; brochure is available)

OPC Inc.
509 West 16th
Cheyenne, WY 82001
(307) 634-3539
(Custom made shoes for M,W, and C)

Bay Orthopedic Shoe Specialties
2712 General Pershing
New Orleans, LA 70115
(504) 899-2132
(Custom and ready-made shoes for M,W, and C; insoles made for sneakers)

Sherman's Shoes
3340 San Gabriel Blvd.
Rosemead, CA 91770
(818) 280-5255
(Ready-made shoes for M and W)

Dey T O Service Corporation
509 5th Avenue
New York,NY 10017
(212) 682-4790
(Custom made shoes for M,W, and C; brochure is available)

Natural Mold Shoe Corporation
49 Lawton Street
New Rochelle, NY 10801
(914) 576-1133
(Custom and ready-made shoes for M and W)

Mathias Boot Makers
20 East 69th Street
New York, NY 10021
(212) 737-3984
(Custom made shoes for M,W, and C)

Hitchcock Shoes, Inc.
165 Beal Street
Hingham, MA 02043
(617) 749-3260
(Mens' shoes only; 100 styles; EEE–EEEEEEWide up to size 13; brochure is available)

Sherman Brothers Men's Shoes:
• 1520 Sansom Street
Philadelphia, PA 19102
(215) 561-4550
• Rosemont Village Mall
1149 Lancaster Ave.
Rosemont, PA 19010
(215) 527-2323
• Corner House Plaza
Rt. 663
Quakertown, PA 18951
(215) 247-7888
(Casual & dress men's shoes sizes 5 1/2-16, AAA-EEE widths)

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CONFERENCE *(Cont. from p. 2)*
hand. After a demonstration of her manual dexterity, conference participants were impressed by the level of coordination Joan has attained since embracing PT.

At the start of her therapy Joan observed patients with much more debilitating problems than her own CMT-related ones. She felt like a cheater who didn't deserve treatment because there were others with worse problems. Joan soon realized the whole purpose of PT was her right to function at her best level. She joined the efforts of PT patients everywhere who seek to improve their quality of life. Joan now strongly advocates PT, explaining that this collaboration between therapist and CMT patient can make a profound difference in the disease's progression.

She urged patients not to accept their diagnosis as a life sentence with periodic returns to the doctor for check-ups on deterioration. The importance of PT is that it can help one deal with the disease and go beyond it. It can liberate your mind, improve your life, and give you the "take-charge" tools that promote being an active patient as opposed to a passive one waiting to waste away.

The NFPMA would like to expand its regional conference program beyond the Philadelphia, New York, and Washington, D.C. areas. Conference discussions are open to the public at no cost and a small fee is charged for lunch at the meeting site. NFPMA funds are used for building rental, speaker stipends, information packets, mailing and registration costs, etc. This program has been vital in spreading the message of CMT patients and organizing them into a stronger patient community, working together for more research into the causes and treatment of Peroneal Muscular Atrophy.

The NFPMA recognizes the great need for regional conferences throughout the United States. This organization would like to take its "traveling road show" and educate both the patients and the public about CMT, but the need for financial support of this program is also great. Each regional conference costs about $10,000 to produce, start to finish. Because of this great expense, the NFPMA welcomes the backing of private foundations and/or community sponsors for its regional conferences.

If you would like to have an NFPMA regional conference in your city, and you can also provide some concrete funding ideas for it, please contact:
Howard K. Shapiro, Ph.D.
NFPMA
University City Science Center
3624 Market Street
Philadelphia, PA 19104
For telephone inquiries:
(215) 664-6010.

FROM THE DESK OF OUR DIRECTOR OF SCIENTIFIC PROGRAMS:

A Fond Farewell to the Official NFPMA Rented Typewriter

The NFPMA wishes to extend a special thanks to Safeguard Business Systems, Inc. of Fort Washington, PA and Diane Freaney, their Vice President and Treasurer, for donation of an IBM XT computer system. We have needed a computer for some time. The equipment recently donated to our program will help us stay in touch with CMT patient families and medical specialists, keep our financial records in order to help "crunch" the raw data from our continuing research studies.

HIGHLIGHTS FROM THE BIG APPLE

During 1985–1986, the NFPMA has sponsored two all day patient family/medical professional conferences and one support group meeting in the New York metropolitan area.

Speakers at these meetings included Fred Gilbert, M.D. (medical geneticist, Mt. Sinai School of Medicine), Carol A. Oatis, P.T., Ph.D. (physical therapist, Philadelphia Institute for Physical Therapy), Nicholas Tzimas, M.D. (orthopedic surgeon, New York University Medical Center), Robert E. Lovelace, M.D. (neurologist, Columbia Presbyterian Medical Center—CPMC), Rosamond Kane, M.D. (orthopedist, CPMC), Anne Lancellotti, P.T. (physical therapist, Rehabilitation Clinic, CPMC), and Howard K. Shapiro, Ph. D. (NFPMA).

Our special thanks to invited speakers for helping out. The NFPMA is now in the process of incorporating a New York State Chapter and expects this to be completed shortly.

For more information on our program in the New York metropolitan area please contact Mrs. Carolyn Redell at (212) 724–3958.

HISTORIC MEETING SCHEDULED FOR NEXT JUNE

The NFPMA in association with the University City Science Center of Philadelphia is sponsoring an international symposium on Charcot-Marie-Tooth Disease. This conference is planned for June 28—July 1, 1987 at Arden House, the Columbia University Conference Center.

This is only the second international research and medical meeting directly focused on CMT disease. In 1976 CMT was the principal topic of the Fourth International Meeting on Neuromuscular Diseases in Marseilles, France.

Seventeen foreign speakers and twenty domestic speakers will participate. In addition, approximately sixty other medical authorities interested in CMT and related topics will be invited to attend.

Only within the past ten years has research on this common disorder shifted to the biochemical level. At least ten laboratories are now doing gene mapping studies on CMT, several other biochemical studies have been completed and several well organized experimental drug therapy studies are either now in progress or have recently been completed.

Reports on these advances have appeared piecemeal over the past decade. The symposium will be the first opportunity for virtually all leading authorities to gather for a comprehensive review of CMT.

In addition to providing a forum for discussion of recent work on CMT, this symposium will serve to stimulate professional interest in the topic and hopefully encourage an increasing number of future studies on this class of genetic disorders.

Our special thanks to **Fidia Pharmaceutical**, an international corporation, for their recent donation of $10,000 towards support of this upcoming research meeting.

THERAPY *(Continued from page 3)*

therapy to patients with CMT. This usually pays for the evaluation and may pay for one visit per month. It may also provide money for orthotic devices (braces) or other devices to improve function.

Physical therapy is an essential part of the treatment of CMT, but finding the right therapist for you requires some research. Paying for physical therapy services depends on your personal insurance, but at least some assistance should be available to you.

Carol A. Oatis, P.T., Ph.D., is a licensed physical therapist and a partner in the Philadelphia Institute for Physical Therapy, has worked with CMT patients for fourteen years, specializing in biomechanics and the feet. Dr. Oatis is involved in several research projects, teaches at Beaver College (PA), and has published a number of articles on various aspects of physical therapy.

GET IN TOUCH

You can keep up to date on our activities and help spread the message of the NFPMA. If this issue was not mailed to you, we invite you to join our mailing list by filling out the form below. If you are already on our list, kindly send the name of anyone else interested in the NFPMA report. Mail this form to:

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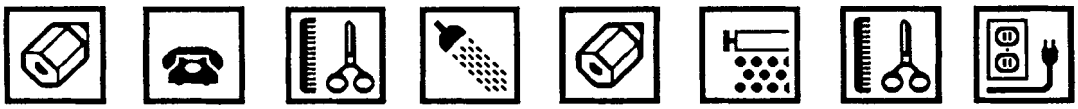

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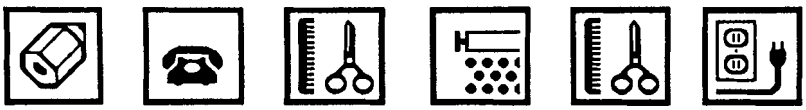

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