

# THE NFPMA REPORT

Spring 1987

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

## International Research Conference to Focus on Charcot-Marie-Tooth Disease

*From Australia to Italy medical scientists interested in Charcot-Marie-Tooth disease are now finishing work on their scheduled presentations at the Second International Conference on Charcot-Marie-Tooth Disorders. CMT disorders are the most common type of inherited neurological disease, affecting perhaps one in every 2,000 to 3,000 Americans. Although as many as 125,000 Americans may have this disorder, almost no public attention has focused on this common class of genetic diseases.*

The Second International Conference on Charcot-Marie-Tooth Disorders will be held this coming June 28th to July 1st at Arden House, the Conference Center of Columbia University. The meeting is funded by the National Foundation for Peroneal Muscular Atrophy and organized in association with Columbia University. To help underwrite expenses for this landmark meeting the NFPMA has secured private support from Fidia Pharmaceutical Corporation, TECA Corporation and major contributions from Mr. Frank T. Crohn, Sr. and the estate of Mr. Seymour Shapiro. Fidia Pharmaceutical Corporation has pursued the development of Cronassial, an experi-

*Continued, Page 2*



The Arden House Conference Center of Columbia University

## CMT Patient vs. U.S. Government

*Job discrimination is a pertinent issue for all disabled people. This newsletter offers J. Michael Springmann's story to encourage CMT patients and to inform employers and medical professionals who deal with these patients. The NFPMA is proud to have assisted Michael in his struggle for fair treatment.*

In September, 1987, J. Michael Springmann is moving to Jeddah, Saudi Arabia. By the time he finishes his 18 month tour of duty as a consular officer for the U.S. State Department there, our government will have spent between \$100,000 and \$200,000 in various training and/or living expenses to keep Michael in the foreign service of his country. In that respect Michael is no different from the 31 other members of his foreign service class. Nine and a half months of instruction in State Department background, language study, and consulate procedures coupled with



salary, moving expenses, housing costs, and hardship differential amount to quite an investment in personnel. To Michael Springmann's tab, however, add substan-

*Continued, Page 5*

### INSIDE:

Get in Touch	6
VCR Tape Rental	6
Rehab Medicine and the CMT Patient	7
Research Update	9
Not-Your-Neighborhood	
Shoe Store	11

*Int'l. Research Conference, Continued  
from Page 1*

imental drug which sometimes is of benefit to patients with peripheral nerve damage. TECA Corporation manufactures some of the state-of-the-art electrodiagnostic equipment now used in electromyography laboratories.

In addition this conference will be partially supported by a grant from the National Institutes of Health. The awarding of a \$15,000 grant from the NIH is itself a notable landmark for those of us working on the NFPMA program. This is the first Federal Government award to our four-year-old program.

Intended as a *workshop* conference, the size of this meeting has been kept small. The meeting will include approximately 40 speakers and an additional 70 attendees who are recognized authorities on topics related to CMT. Dr. Peter James Dyck of the Mayo Foundation will be giving the keynote lecture for this conference. In organizing this meeting, Dr. Howard K. Shapiro of the NFPMA has worked closely with Dr. Robert E. Lovelace of the Neurological Institute, Columbia University.

Surrounded by state forest reserves 40 miles up the Hudson River from New York City, Arden House is an ideal location for a workshop conference of this kind. Prior to being donated by Ambassador W. Averell Harriman and E. Roland Harriman to Columbia University, this 143 room country estate was the ancestral home of the Harriman family. Opened in 1950 as the nation's first educational conference center, this unique facility continues to fulfill its role as a congenial site for international conferences on a wide variety of issues.



The first international meeting to specifically address CMT disorders was the Fourth International Meeting on Neuromuscular Diseases, held in Marseilles, France in 1976. This meeting, organized by Dr. Georges T. Serratrice, dealt primarily with clinical and electrophysiological aspects of CMT, and also included discussions on a related topic, the scapulo-peroneal syndromes. Little biochemical research had been done on CMT at that time. The Marseilles conference proceedings were published in a text entitled Peroneal Atrophies and Related Disorders (Masson Publishing USA, New York, 1979).

Fortunately, the past decade has seen an unprecedented increase in attention to CMT as a biochemical research topic. Ten years ago the most heated professional debates on this disease may have been related to clinical definitions which describe the physical status of patients. These issues included questions relating to age of clinical onset, disease progression, physical degeneration of peripheral nerve fibers and other clinical aspects of CMT. Many of these topics will also be discussed at the Arden House meeting, as additional information on them is now available.



*Continued, Page 3*

*Continued from Page 2*

However, in recent years the focus of CMT medical scientists has shifted towards experimental drug trials and biochemical laboratory studies. Some of these studies have been financially supported by the National Institutes of Health and the Muscular Dystrophy Association (USA). Government health agencies and muscular dystrophy programs in other countries have also funded some of this recent work. A new entry on the CMT research scene, the National Foundation for Peroneal Muscular Atrophy has supported the research studies of Dr. Shapiro, our scientific program director, for the past three years. Dr. Shapiro's studies at the University of Pennsylvania Medical School, discussed at some length in our previous edition of the *NFPMA REPORT*, deal with procedures for comprehensive physiological screening of CMT patients.

#### **One basic question: why have a meeting at all?**

As plans for this important conference approach completion, some observers may ask the most fundamental question: what is the value of such a meeting? There are several good reasons for organizing this program. Such an event focuses attention on CMT within the medical community and brings this issue to the attention of the general public. As CMT patients continue to discover, we live in an age when even many physicians have still not heard of this common type of genetic disorder. Increased awareness of this problem within the medical community translates in the long run into better service of the patient needs and recognition of this problem as a legitimate research topic.

Although international neurology meetings are occurring on a regular basis, CMT is rarely a priority issue, and is never the issue of first importance. Almost every leading authority on CMT will be at the Arden House conference, discussing recently completed work and future plans. Some of the research studies to be presented at this con-



ference are so new that their authors have requested that abstracts of their lectures now in our hands not be released until shortly before the meeting. This gives these investigators time to submit reports of these projects to medical journals.

This meeting is also our way of acknowledging the efforts of concerned medical professionals. These people, after all, have pursued a largely thankless task. Each in his/her own way has dedicated himself/herself to learning more about a disease which is unknown to the general public, has a name which stimulates more jokes than interest, and has proven quite resistant to successful research. At their home institutions each of these medical authorities find few of their colleagues sharing interest

in or knowledge of CMT. Yet during the past decade much progress has been made in understanding the biological mechanisms which underlie CMT.

The Arden House CMT meeting is organized into five sessions, each of which includes from seven to ten lectures. Session topics include: clinical studies and pathological alterations, clinical neurophysiology and clinical heterogeneity, axonal and Schwann cell metabolism, linkage studies and genetic heterogeneity, and metabolic studies on CMT syndromes and experimental drug trials. Interspersed in the formal lecture program are panel discussion breaks. Such informal discussions are an important part of a meeting of this kind.

*Continued, Page 4*

Continued from Page 3

**Three of the lecturers at this conference have a special interest in this topic, as they are also CMT patients**

Three of the medical scientists presenting work at Arden House have personal familiarity with CMT, as they too are patients. These are Dr. Lowell L. Williams of the Children's Hospital of Columbus, Ohio, Dr. James R. Lupski of the Baylor College of Medicine and Dr. Howard K. Shapiro of our own program.

Dr. Williams may be described as an example of the *CMT over-achiever* phenomenon. She is a practicing pediatrician and mother of five children. That would be enough to keep most people busy. However, Dr. Williams has also organized her own multidisciplinary research program on CMT. This includes research on fatty acid metabolism, the immunologic status of CMT patients and gene mapping studies on CMT. Dr. Williams is the only speaker scheduled to give three research lectures at the Arden House CMT conference.

Dr. Lupski has both M.D. and Ph.D. doctorate degrees (New York University). His undergraduate studies included work at the prestigious Cold Spring Harbor Laboratories. He is now a Research Assistant Professor of Molecular Genetics and Pediatrics at Baylor Medical Center (Houston, TX). It is now well established that some CMT families showing autosomal dominant inheritance have a genetic defect on chromosome one. For this pattern of inheritance approximately half the children of a CMT patient will show the disease, but the genetic defect is not on the X chromosome. However, the majority of autosomal dominant CMT families studied so far do not show evidence of a chromosome one defect. At the moment molecular geneticists are stymied on this question.

Dr. Lupski is pursuing a research opportunity which may answer this

question, a least partly. He is studying an unusual autosomal dominant family where CMT patients also have a second genetic defect, von Willebrand disease. This is a blood clotting factor disorder which also shows autosomal dominant inheritance. Earlier studies have shown that von Willebrand disease results from a genetic defect on chromosome twelve. Dr. Lupski is applying the latest techniques of molecular genetics to determine if CMT in this family also results from a problem on chromosome twelve. Patients in this unusual family may be missing a piece of deoxyribonucleic acid (DNA) from two neighboring genes.

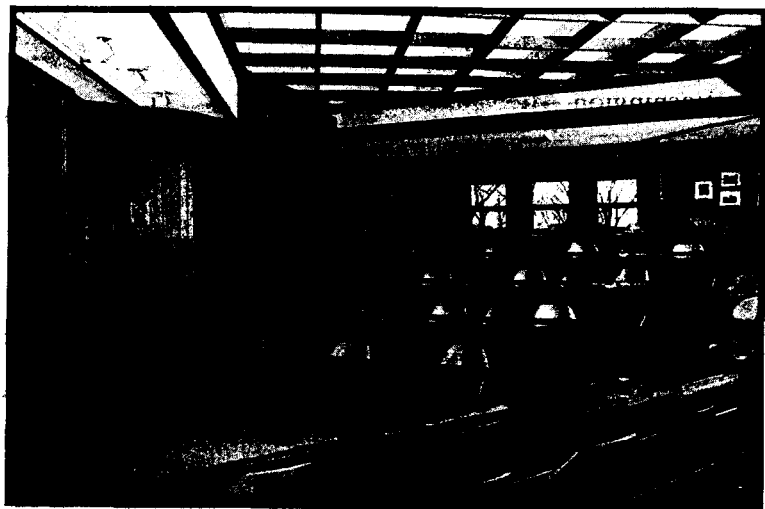
### Other Special Attendees

Dr. Philip F. Chance is a pediatric neurologist who also does gene mapping studies of CMT families. This is the kind of laboratory work that will first identify the exact loca-

tion of the exact location of the first medical scientist to do extensive gene mapping work on CMT. Dr. Chance is currently the recipient of the largest grant ever awarded by the National Institutes of Health for research on CMT.

Another special lecturer at the Second International Conference on Charcot-Marie-Tooth Disorders will be Lyn Griffiths of the University of Sydney (Australia). Ms. Griffiths is a graduate student now finishing her Ph.D. studies in the laboratory of Dr. Garth A. Nicholson. Like Dr. Chance, Ms. Griffiths has done extensive gene mapping studies on CMT, and will give the opening lecture on this topic. She is the first person to get a doctorate degree studying CMT since the medical school studies of Dr. H. H. Tooth a century ago.

Plans for the Second International conference on Charcot-Marie-Tooth Disorders have proceeded



tions of CMT genetic defects on chromosomes and then lead to isolation and further characterization of the genes. Chromosomes are the strands of DNA (deoxyribonucleic acid) present in each human cell which bear the characteristics (genes) that we inherit from our parents. Each of the 23 pairs of human chromosomes has several thousand genes in its structure, so identification of the exact site of a coding error in one gene is a formidable problem. Dr. Chance received his research training in the laboratory of Dr. Thomas D. Bird (another conference attendee), the

remarkably well. We expect a program unlike any that has come before. The proceedings of this meeting will, in turn, serve as the basis for a subsequent book. It is true that the riddle of CMT has been a tough nut to crack, but successful work is now underway in many laboratories. The past decade has seen the first comprehensive biochemical research on CMT. The next *NFPMA REPORT* will review the proceedings of this important conference in terms of scientific content and how current work may affect and serve CMT patients.

*CMT Patient vs. U.S. Government,  
Continued from Page 1*

tial legal and administrative expenses — incurred by the United States Government defending itself against an Equal Employment Opportunity complaint and a lawsuit. Lawyers, doctors, investigators and administrative staff were occupied for two years because of a misunderstanding about the nature of Charcot-Marie-Tooth disease.

Michael Springmann has had the kind of career that most people only dream about. With a B.S. from the Foreign Service School of Georgetown University and a graduate degree in International Affairs, from 1977 to 1980 he was assigned to Stuttgart, West Germany in a State Department — Commerce Department exchange program. One of his major functions was to visit companies and provide U.S. government service in trade relations. This assignment involved plenty of walking, driving, and climbing stairs. After a brief return to Washington, Michael was off again to lead a similar life of office work and client visitation in New Delhi, India.

Back in D.C. in 1982 with the Department of Commerce, Michael became restless for more travel. At this time he decided to seek a career with the State Department, competing with 15,000 other applicants for 200 available positions. Successful at both the written and oral exams, Michael acquired his security clearance and proceeded to his medical screening.

At that examination Michael Springmann was told, "There's something wrong with you, but we don't know what it is." An EMG (electromyogram) and further examination by a neurologist (not a State Department employee) revealed that Michael has Charcot-Marie-Tooth disease. So at 40 years

old Michael was first diagnosed as a CMT patient.

Previously unaware of CMT and its neurological basis, Michael only thought he had "lousy feet" with hammertoes and very high arches causing callouses and corns. Sprained ankles in graduate school caused him to seek advice from an orthopedic surgeon. This doctor did not recognize CMT, but nevertheless did some corrective surgery on Michael's feet and ankles.

Further complicating Michael's situation is an absence of family history because he was adopted. At this writing he still awaits an October, 1986 promise from U.S. District Court to deliver a decision about releasing his adoption record. In the meantime, CMT may be present in Michael's eleven-year-old daughter.

Although Peroneal Muscular Atrophy and CMT disease were new words to both Michael's vocabulary and life, his first assurance from the diagnosing neurologist was that CMT would not be a factor in his ability to participate in the foreign service. Later his case was reviewed by the Medical Review Committee of the State Department. After examining Michael's paperwork (misleading



Happy to be training for foreign service with the U.S. State Department, Michael Springmann spoke with us at his Washington, D.C. residence.

and badly prepared by a consulting neurologist at Georgetown University Hospital), this group denied his medical clearance. Michael was shocked to learn that the neurologist who first reassured him had later submitted a negative report stating that Michael could only serve where there are elevators and adequate public transportation.

Now Michael plunged himself into obtaining as much knowledge about his condition as he could. In his efforts to counter the consultants' claims, he consulted with and received aid from Dr. David Pleasure of the University of Pennsylvania and Dr. Ruediger Kratz, referred by the Muscular Dystrophy Association. He approached various other organizations who work on behalf of the handicapped, but was disappointed by a lack of support from them. It was his inquiry at the National Institutes of Health that brought the NFPMA to Michael's attention and put him in touch with Dr. Shapiro, our Director of Scientific Program, who spoke with Michael about CMT and encouraged him in his efforts.

About two months later the Civil Rights Division of the Department of Justice contacted Dr. Shapiro seeking information about CMT for a case. They asked Dr. Shapiro for a written statement about CMT-related physical disability and job performance.

In his letter, Dr. Shapiro acknowledged his familiarity with Michael Springmann's case. In drafting a statement Dr. Shapiro understood that job discrimination is a potential issue for any CMT patient. A key question is how much an employer should expect from a CMT employee. Dr. Shapiro's answer was that these patients perform as well as their co-workers, sometimes better. In reviewing examples of professionally successful CMT patients, Dr. Shapiro referred to four physicians who are CMT patients, five CMT lawyers on the NFPMA mailing list and a special education teacher who received the 1984 Teacher of the Year Award

*Continued, Page 6*

from the National Society for Autistic Children and Adults. Dr. Shapiro noted that "in spite of their physical disabilities CMT patients engage successfully in a wide range of professional activities and frequently excel in their work."

Energetically gathering information and testimony such as Dr. Shapiro's, Michael first tried to appeal his case through the appropriate steps within the State Department under the guidelines of the Civil Rights Act and the Equal Employment Act.

After a year of unsuccessful negotiations with the Department of State, Michael sued the United States and Secretary of State George Schultz in U. S. District Court in Washington, D.C. At that time another consulting neurologist, Dr. John Griffin of Johns Hopkins University Hospital, was chosen by the Department of Justice to see Michael. Dr. Griffin later submitted a report stating that there was no reason why Michael Springmann could not be hired for foreign service. He reiterated much of what Michael had already learned about CMT. This disease is slowly progressive, individual to personal cases, and will not deter Michael from doing a fine job wherever he is assigned. There is also no need for Michael to be medically evacuated for treatment for CMT.

Happily, after Dr. Griffin's review the State Department withdrew and settled out of court. In November, 1986, nearly two years after his medical clearance was denied, Michael Springmann entered into a State Department foreign service training class. He is presently in the midst of a six month class in Arabic and is looking forward to the overseas assignment he fought so hard to win.

## VCR Tape Rental

The NFPMA will rent 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 hour, 15 min.) rent for \$10, and the double lecture tape (2 1/2 hrs.) rents for \$15. The rental fee includes prepaid return postage. To order a tape, fill out our *Keep in Touch* form and send it with a check or money order to the NFPMA, University City Science Center, 3624 Market Street, Philadelphia, PA 19104.

### 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**. You can also use this form for VCR tape rentals. Mail this form to:

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Tell us about yourself (optional):

CMT patient

Interested supporter

Medical professional

CMT family member

Research scientist

Other \_\_\_\_\_

## 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 (curvature of the spine).
- . . . 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.

## Rehab Medicine and the CMT Patient: AN UNRECOGNIZED OPTION



*"If there is a practical specialty in medicine it is physical rehabilitation, whose real function is to keep people functioning. The parameters are as large as anyone's*

*imagination. Because it doesn't exist doesn't mean it can't be done." These optimistic words spoken by Dr. Robert J. Krebs III characterize his enlightened and enthusiastic approach to the specialized field of rehabilitative medicine. The following excerpts from a recent NFPMA interview with Dr. Krebs confirm our belief that CMT patients can and should take a proactive approach to this disease.*

**Q:** What is rehabilitative medicine?

**A:** Rehabilitative medicine is a specialty where the primary goal is to return people to functional existence. This is accomplished in two ways. First, through the restoration of previous functional ability by providing physical therapy exercises. Second, through the

adaptation of different mechanical devices (for example bracing) to accomplish lost function.

**Q:** What type of training does a Rehabilitative Specialist receive?

**A:** To obtain Board Certification a medical physician enters a two year residency program in Rehabilitation Medicine. He is then required to pass both oral and written boards.

**Q:** How frequently do you see people with CMT?

**A:** Unfortunately rehabilitation is not part of neurological training, and because of this most neurologists do not send their CMT patients for a physical rehabilitation evaluation. Most of the CMT patients that we do see come to rehabilitation evaluation as a result of other problems. A patient is often not even aware that he has CMT, and in the course of an evaluation it is determined that CMT is a strong possibility. At this point we

would send this patient to a neurologist and recommend that the entire family be tested.

**Q:** What would you describe as the most common type of physical rehabilitation programs used on CMT patients?

**A:** In dealing with CMT there are three initial areas to examine: prevention of bone fracture, maintaining muscle strength and muscle endurance, and assisting function, if function is lost. In most cases rehabilitative therapy would concentrate on building endurance. Endurance is critical to a CMT patient because it helps a muscle maintain function day to day. **Endurance exercises** are low in weight, high in repetition. It is extremely important in any exercise program to avoid fatigue. This is why it is vital to have medical supervision to monitor and set parameters.

**Q:** How does someone go about finding a specialist in rehabilitative medicine?

**A:** In most areas you can call a local hospital and ask if they have a specialist in the Department of Rehabilitation. Often community hospitals have facilities and they are easy to reach. If a patient lives far away he can come in for an evaluation and then return home with a prescription which can be followed by a certified physical therapist.

**Q:** What is the procedure for a patient coming in for the first time?

**A:** First the rehabilitation specialist must be sure that the diagnosis is correct. A complete neurological work-up by a competent specialist must be done to determine that what appears to be CMT is. Then the patient would see the rehab specialist for an extensive examination. This would include a discussion of the patient's goals and expectations and what the patient needs to function better in

*Continued, Page 8*



his life. The next step in the process is testing out these goals in a supervised setting at a physical therapy facility. An occupational therapy evaluation is also done at this time. Patients with more advanced function lost will be examined by a team consisting of an orthopedist, prosthetist and podiatrist.

**Q:** What is the time investment for this procedure?

**A:** A patient can expect to spend an hour for a full evaluation history. Then three to six one-hour sessions of therapy to set the parameters (this will depend on how involved the patient is). These sessions are scheduled three times a week for two weeks. At this point the patient is fully prepared to carry out the prescribed exercises. A follow-up is needed every six months minimally. This depends on the progression of the disease.

**Q:** What are the costs incurred?

**A:** \$100 to \$150 for the physician's consultation fee. Physical therapy runs from about \$50 to \$70 per session. Major medical or group insurance usually covers 80% to 85% of the cost.

**Q:** Does an HMO cover physical rehabilitation costs?

**A:** Not all HMOs are what they appear to be. Some are mechanisms to make money and offer very little. A true HMO should offer physical therapy coverage. If your plan does not, I would see about changing into another plan when the opportunity presents itself. Any plan worth its salt should offer this coverage and all the good ones do.

**Q:** Are there any notable success stories on CMT patients and physical therapy?

**A:** I have not seen any good studies where we have taken people in the early stages of the disease, classified them from a neurological standpoint, put them into a really good comprehensive physical rehabilitation program and then followed them over a long period of time. Often people get lost after the first evaluation or fall off in rehabilitation. This is partly our fault. We don't know what happens to them in five or ten years. It would be great if, through the NFPMA you could get across to people that this is not a one-time thing. It has to be a long-term maintenance program.



NFPMA staff discusses hand mobility with Dr. Krebs. Left to right: Mary Tracy, Lydia Brennan, Dr. Krebs and Karol Hitt.

**Q:** Have you ever used electrical stimulation in a therapy program?

**A:** Electrical stimulation is debatable from an academic viewpoint. This procedure tends to fatigue muscles too soon. A CMT patient usually has low muscle reserve, his muscles are working at maximum capacity. If the disease is at an early stage and we wanted to help utilize muscle that is not normally utilized then it might be worthwhile. I think that it is an interesting area for the future, but more research is needed.

**Q:** What are good exercises for a CMT patient in a workout club?

**A:** Anyone who walks into a health club and randomly exercises

on equipment is at risk. A CMT patient is at higher risk. The proper exercises should be prescribed by a rehabilitation specialist and tested out in therapy. This way a patient learns what he is capable of doing, what his reserves are, the types of exercises that should be performed and the maximum amount of weights that can be used. With this knowledge a CMT patient can go to a health club knowing his limits and goals.

**Q:** Is jogging too traumatic for the CMT patient?

**A:** Jogging is traumatic for most

people. I think there are other forms of exercise that will accomplish what you want. Walking at a quick pace is 85% as efficient as jogging and far easier on the joints. Swimming and bicycling are both excellent ways to keep in shape. I would not recommend jogging to anyone with problems, particularly CMT patients.

**Q:** Does CMT affect the heart?

**A:** I don't see this as a problem in the majority of CMT patients. What may happen is that in certain families, neighboring genetic loci travel together. By coincidence, one will carry a congenital heart problem and the other CMT.



**Q:** It seems that CMT patients go quickly downhill after they reach 60. Can you explain why this happens and how it can be controlled?

**A:** As age sets in, muscles lose some of their strength. A person without CMT has enough muscle reserves to replace what is lost. CMT patients do not have this reserve, they use it all to function. When the natural process of aging takes away this reserve they have nothing to replace it. They lose function, thus becoming disabled by the aging process. This loss of function can be controlled by keeping the reserves up. Muscles must be kept to their peak of endurance and strength.

**Q:** Any help at all for the 70 year-old patient in a wheelchair?

**A:** Unfortunately it often gets to the point of no return. The atrophy occurs, the muscles go and that's it. Once they go, they're gone.

**Q:** Do you have any special thoughts on dealing with children?

**A:** Parents should seek out people who specialize in pediatric rehabilitation (an even more limited field). Also find therapists who specialize in pediatric therapy. An environment where the children feel comfortable will go a long way in assuring a successful treatment. In the case where a child is interested in sports, a rehabilitation specialist can often prescribe splints or special supports to facilitate better movement.

*Robert J. Krebs III, M.D., M.P.H., a Board Certified Rehabilitation Specialist, is Chairman of the Physical Medicine and Rehabilitation Departments of Brandywine, Chester County, and Delaware County Memorial Hospitals in Pennsylvania. Dr. Krebs, an Assistant Clinical Professor at the University of Pennsylvania School of Medicine, has written several articles on rehabilitation and gives frequent lectures on the various aspects of this field.*

Our special thanks to the National Foundation for Jewish Genetic Diseases (NFJGD) and its President, Mr. George Crohn, for a 1987 grant of \$4,000 to support the research studies of Dr. Shapiro on familial dysautonomia. Familial Dysautonomia is a rare genetic disorder which, like CMT, affects nerve fibers. Dr. Shapiro's study on serum lipids (fats) in familial dysautonomia patients, conducted in collaboration with Dr. David J. Prescott of Bryn Mawr College, is an off-shoot of an earlier study on CMT serum lipids. The current NFJGD grant follows a 1986 NFJGD grant of \$3,500 for initiation of this work.

## Research Update

Since publication of our previous edition of the *NFPMA REPORT* we have received questions from several CMT patients as to how they might alter their diets to avoid furan aldehyde products. Furan aldehydes may be generated from certain sugars during the cooking of food. At this time, we are not advising CMT patients to alter their diets.

In recent months we have received confirmation from an independent testing laboratory (Meridian Instruments) regarding our findings on the CMT family described in our previous edition. These CMT patients do indeed have a problem metabolizing furan aldehyde products, which may be the basis of their disease. However, we also sent Meridian Laboratories urine samples from CMT patients in a second family group and these patients showed normal, or near normal, abilities to metabolize furan aldehyde products. This is not especially surprising, since we already know that there are several different genetic varieties of CMT. These findings are quite new and not yet reported in the medical literature as full papers. This will be done shortly.

Although we have some original, interesting findings from this work, the testing procedures are experimental and expensive. We will be extending these studies as our funds permit, and we hope these results will hold true for many CMT patients. However, any general

recommendations concerning alteration of CMT patient diets must await additional laboratory studies. Our latest findings will be presented at the Second International Conference on Charcot-Marie-Tooth Disorders and we will, of course, continue to keep our readers posted on the development of this work.

— Howard K. Shapiro, Ph.D.  
NFPMA Scientific Program Director

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## To Our Readers...

The response to the first *NFPMA REPORT* has been very positive. We have heard from patients, their families, and medical professionals encouraging us with comments and suggestions. Some have asked how to financially help the NFPMA, and that is very easy. Your contributions are not only greatly appreciated, but very necessary. We are a tax-exempt, non-profit medical foundation and exist on voluntary contributions. Patient support, public education programs, the *NFPMA REPORT*, Dr. Shapiro's research, and special activities such as the Second International Conference on CMT Disorders are **only** possible because of individual, group and corporate donations. All donations are gratefully accepted, and checks can be made out to the National Foundation for Peroneal Muscular Atrophy (or to NFPMA) and sent to the return address on the cover. With increased funding we will expand our CMT patient/family, medical/pro-

fessional conferences and support groups to other parts of the country. Please write a check today and help us to continue our work.

You may want to consider remembering the NFPMA in your will. The hereditary nature of CMT makes this a particularly appropriate way for patient families to leave a living legacy. (Currently part of the Arden House International Research Conference is being funded by a thoughtful bequest.) If your employer has a matching gifts program, advise them of your donation so that we may benefit from their generosity also. Finally, many fraternal and civic organizations have discretionary funds and donate regularly to worthy causes. Spread the word to them that we are here and deserving of their consideration. Please help us; we need you.

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## **NFPMA REMEMBERANCES**

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Your gift to the NFPMA can honor a living person or the memory of a friend or loved one. Acknowledgement cards sent *in honor of* or *in memory of* will be mailed by the NFPMA 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 NFPMA by completing the form below and mailing it with your check to **NFPMA, University City Science Center, 3624 Market Street, Philadelphia, PA 19104.**

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Phone Number: \_\_\_\_\_

### Memorial Gift

In memory of: (name of deceased)  
\_\_\_\_\_  
\_\_\_\_\_

Send acknowledgement to:

Name: \_\_\_\_\_

Address: \_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_

### Honorary Gift

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

Send acknowledgement to:

Name: \_\_\_\_\_

Address: \_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_

Occasion:

Birthday  Holiday \_\_\_\_\_

Wedding  Thank-you

Anniversary  Other \_\_\_\_\_

Amount Enclosed: \_\_\_\_\_

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

# 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 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 of special shoes. Please let us know if you have additional shoe ideas.

## The following are retail stores:

Peterson Shoe Store  
209 East Main St.  
Anoka, MN 55303

Kates Brothers  
188 Jefferson St.  
Passaic, NJ 07055  
(201) 473-7318

Francis Bacon  
206 N. Liberty St.  
Baltimore, MD 21201  
(301) 727-3775

Bloch and Co.  
1014 West 38th St.  
Austin, TX 78705  
(512) 458-5115

Bogers Shoes  
1014 Park St.  
Jacksonville, FL 32204

Henderson Shoes  
280 Sunrise Hwy.  
Rockville Center, NY 11570

Windham's Shoes  
12419 S. Hawthorne Blvd.  
Hawthorne, CA 90250  
(213) 675-8106

McMahan Shoes, Inc.  
505 Courtland St., N.E.  
Atlanta, GA 30308  
(404) 874-3831

Brown Shoes, Inc.  
2108 Tremont Center  
Columbus, Ohio 43221  
(614) 468-4477

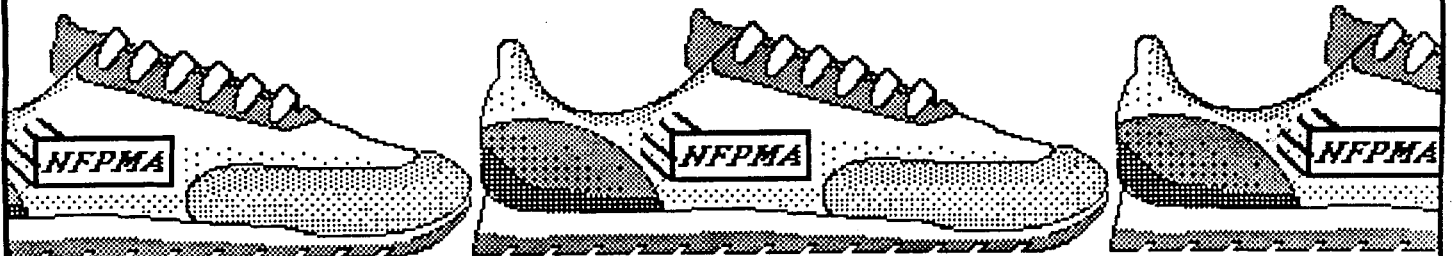
Jerry's Style-Opedic Shoes  
1753 East 17th St.  
Denver, CO 80218  
(303) 333-8877

Reicken's Shoes  
401 N. Green River Rd.  
Evansville, IN 47715  
(812) 476-8006

Eneslow Comfort Shoes  
697 Avenue of the Americas  
(corner at 22nd. St.)  
New York, NY 10010-5105  
(212) 929-1810  
(7 other NYC locations,  
and also in Tamarac, FL)

**Manufacturers** — This company will answer calls and refer the caller to the nearest store carrying their shoes.

P.W. Minor & Son, Inc.  
3 Treadway Avenue  
P.O. Box 678  
Batavia, NY 14020  
(716) 343-1500



**GADGET INFO** — *So you almost strangled yourself yesterday getting the neck button on your shirt closed and last week it took twenty minutes to open a jar of pickles. Well, take heart. We are seeking out information sources for household products to help the handicapped. If you have any favorite gadget sources, please let us know. To obtain gadget information try contacting:*

**Bruce Medical Supply**

Dept. CMT  
411 Waverly Oaks Rd.  
Waltham, MA 02154  
(800) 342-8955



**Independent Living Aids, Inc.**

11 Commercial Court  
Plainview, NY 11803  
(800) 262-7827



**Comfortably Yours  
(Aids for Easier Living)**

52 West Hunter Avenue  
Maywood, NJ 07607  
(201) 368-0400



**Enrichments, Inc.**

P.O. Box 579  
145 Tower Drive  
Hinsdale, IL 60521  
(800) 343-9742

**ATTENTION MEDICAL SPECIALISTS**

**—CALL FOR RESEARCH SAMPLES—**

The successful metabolic screening studies carried out by the NFPMA are now being extended to include patients representing two other genetic neuromuscular diseases: Huntington's disease and Duchenne muscular dystrophy. The technology used in this program (gas chromatography/mass spectrometry) is the most powerful metabolic screening tool available, yielding a library of physiologic data on each patient. Extending our metabolic screening studies on CMT to include work on related diseases will provide important new information. Patient participation in this research is simple—all we need is one urine sample from each donor. Medical specialists aware of HD or DMD patients willing to participate in this study should contact our Director of Scientific Program for additional information:

Howard K. Shapiro, Ph.D.  
National Foundation for  
Peroneal Muscular Atrophy  
University City Science Center  
3624 Market Street  
Philadelphia, PA 19104  
(215) 664-6010

**THE NFPMA REPORT**

*information on Charcot-Marie-Tooth disease from*  
**National Foundation for Peroneal Muscular Atrophy**

University City Science Center  
3624 Market Street  
Philadelphia, PA 19104

Non-Profit Org.  
U.S. Postage Pd.  
Philadelphia, PA  
Permit No. 3633

TO: