

THE NFPMA REPORT

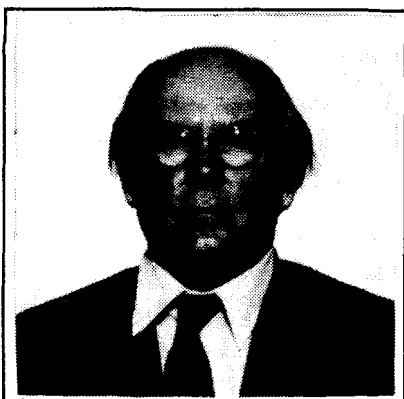
In honor of Henry P. Eldredge, this issue is partially funded by the Rotary Club of East Greenwich, RI

Winter 1989

Vol.3/No.1

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

DR. PETER JAMES DYCK DISCUSSES CMT



The following article is excerpted from the keynote address Dr. Peter James Dyck gave at the Second International Conference on Charcot-Marie-Tooth disorders in June, 1987. The conference was sponsored by the NFPMA and Columbia University. (See The NFPMA Report, Vol. 1, #3.) Dr. Dyck, a world recognized authority on CMT, is a professor of neurology, a researcher, and a clinician at Mayo Medical School, Rochester, Minnesota. He has authored

or co-authored 290 clinical and research articles. Since Dr. Dyck's audience primarily included research scientists, much of his speech was of a technical nature. However, parts of it were of such a nature that we felt every CMT patient should have the opportunity to benefit from Dr. Dyck's insights and advice to physicians and patients.

My interest in inherited neuropathy began in 1962. I began to see patients in a valley of the Mississippi River, the Zumbro River, which flows into Lake Pepin. This kindred lived in the small villages and farms of that area and it turned out that investigators from the University of Minnesota had done genetic studies in this region some fifty years earlier.

I pursued Ed Lambert's discovery of low nerve conduction in inherited neuropathy. We now know much more about inherited neuropathy than we did twenty-five years ago. Some of you here have played important roles in these discoveries. Unfortunately much more remains to be discovered about inherited neuropathy. When we began our studies the question was why are conduction velocities low in

inherited neuropathy? Lambert had made the observation that they sometimes were low, and secondly that they might serve as a marker of Charcot-Marie-Tooth syndrome. We asked a series of questions. Why was nerve conduction low? Did all kindreds with peroneal muscular atrophy have such low nerve conduction? Are the nerves enlarged in peroneal muscular atrophy? Why? Since nerve has fibers of different functional and size classes, which classes were especially vulnerable to disease? Was the neuron, or the axon, or the

(continued next page)

CMT Conference Slated for September

The NFPMA and the Alfred I. Du Pont Institute of Wilmington, DE, announce the sponsorship of a CMT professional/patient conference on September 8 and 9, 1989. The conference will be held at the A. I. Du Pont Institute. Friday's program will be directed to medical professionals and Saturday's topics will be geared to CMT patients and their families. Details will follow in our next issue.

INSIDE:

PATIENT PROFILE.....	P. 3
ASK the DOCTOR.....	P. 6
GROUP NOTES.....	P. 7
VCR TAPE RENTAL.....	P. 11
GET in TOUCH.....	P. 11

Schwann cell, or the myelin selectively involved? What were the three dimensional alterations along the nerve fiber? We also asked what was the metabolic abnormality in these diseases? Were the syndromes with peroneal muscular atrophy distinctly different? Were they from different mutant genes? What was the role of environmental factors? What were the gene loci (chromosome locations)? Are there specific treatments in these disorders? The reason for listing these questions is to show that progress has been made in the last twenty-five years. We have answers for many of these questions.

Since I began my studies in inherited neuropathy we have learned a lot. We know that peroneal muscular atrophy is not one disease but several diseases. We have learned a considerable amount about how the disorders can be detected and characterized. The disorders known as Charcot-Marie-Tooth syndrome may also be referred to as forms of Hereditary Motor and Sensory Neuropathy, or HMSN. Some are directly inherited from an affected parent to one-half of the children (dominantly inherited). Others are recessively inherited (25% of the children from unaffected parents) and still others are inherited (sons) from an unaffected mother (sex-linked). There are different disorders even within these different inheritance patterns. Low nerve conductions are a useful marker for some disorders but not for others.

The approximate chromosome and gene localization is known for two varieties of HMSN, HMSN-Ib and sex-linked HMSN.

We now have some understanding of the structural changes in nerve which account for the clinical symptoms and abnormality of nerve conduction. In HMSN-I all classes of nerve fibers are affected. There is an abnormality of redistribution of enzymes and other macromolecules along the length of the fiber. The nerve fiber appears to develop normally, then prematurely begins to atrophy. This atrophic condition appears to begin in the feet and legs. With atrophy there is myelin remodeling. Repeated de- and re-myelination is involved in developing a microscopic abnormality called the onion-bulb formation and enlargement of the nerve. Disturbed electrical phenomena are associated with fiber atrophy and myelin re-modeling.

Low nerve conductions, in general,

relate to the severity of the clinical deficits. The evidence for myelin remodeling and for the occurrence of secondary demyelination also came from studies of uremic neuropathy, Friedreich's Ataxia (a recessively inherited disorder also associated with peroneal muscular atrophy) and an experimental study of the nerves above the site of the amputation of the legs in cat and man.

Several lines of evidence convinced us that demyelination (breakdown of the fat and protein insulation of a nerve fiber) could, in some cases, be due to axonal atrophy. The reasons were: (1) demyelination was clustered on certain fibers—presumably those with atrophic axons, (2) demyelination was more frequent in more proximal aspects of nerve—distally fibers had undergone degeneration, and (3) in transverse sections of nerve the caliber of axons relative to myelin thickness was decreased as com-

(continued, page 4)



MEDICAL ALERT

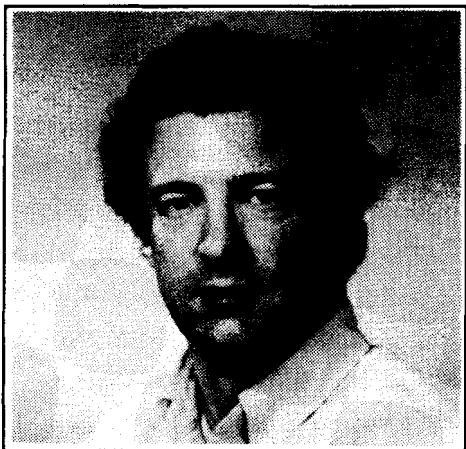
Certain Drugs Toxic to the Peripheral Nervous System

This is a list of neurotoxic drugs which could be harmful to the CMT patient. Before taking any medication, **alert your physician to your specific medical history.**

Adriamycin
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, Macro-dantin)
Nitrous Oxide (chronic repeated inhalation)
Penicillin (Large IV doses only)
Pyridoxine (Vitamin B⁶)
Vincristine

Patient Profile — Steve Gulick



The following article is reprinted with permission from The Annual Report of the Pennsylvania College of Podiatric Medicine (PCPM). Located in Philadelphia, the college is dedicated, "To create, transmit, and conserve knowledge of the foot, and its role as an integral part of total health care."

Unlike other actors, Steve Gulick cannot stand before an audience and forget himself, completely absorbed in his performance. The 40-year-old mime and singer suffers from a degenerative muscle disease known as Charcot- Marie-Tooth Syndrome (CMT), which affects his legs. On stage Steve must focus on how well his legs are functioning, so as not to trip or fall.

Problems with Steve's legs began in high school when he experienced difficulty walking and running. At the time Steve's mother attributed these problems to a bout of measles and encouraged Steve to use his legs more, believing physical activity would help him regain his strength.

Steve actively participated in sports

throughout high school, but in his freshman year of college, concern and even fear set in. "I knew something was wrong when I couldn't pass the running portion of the physical fitness test. At the time, I thought perhaps asthma was to blame," explained Steve.

Over the next several years, Steve's concern grew. "By 1974, I couldn't even jog across the street— my knees would buckle." Finally in 1979, Steve was diagnosed as having CMT.

The prognosis was discouraging since no one was able to present hope for a cure or even improvement. Finally, Steve heard about a study being done at the Pennsylvania College of Podiatric Medicine pertaining to peroneal muscular atrophy, a disorder which encompasses CMT.

Eager to participate in a research project that would help to improve the

treatment of CMT sufferers, Steve agreed to join the research program currently underway at PCPM. He began working with Gilbert A. Hice, DPM, associate professor in the Department of Podiatric Orthopedics, utilizing electrical muscle stimulation (EMS) which is designed to improve the function and physiology of muscles. According to Dr. Hice, the disease Steve suffers from is hereditary and can affect any muscle group in the human body. At present, there is no known cure for CMT, nor do any forms of treatment including EMS guarantee improvement.

Despite the uncertainty, Dr. Hice is optimistic about electrical muscle stimulation, and his persistent work has added another vital element to the healing process— hope. "Since my treatment began, there are clear indications that it's having some effect," Steve said. "Recently while

(continued next page)



Steve Gulick

(continued from page 3)

performing, I noticed it was much easier to stand than it had been for a long time. My lifestyle is the same. I still have to balance myself so my knees don't collapse; I can't just decide to go outside for a long walk, and if I climb steps, there has to be a railing. But since I've been treated at PCPM, I feel more activity in my calf muscles."

Dr. Hice will modify the techniques used to treat Steve based on his progress and that of other subjects who are participating in current CMT research. "While EMS works to improve the physiology in Steve's leg muscles, another method we're using, functional electrical stimulation (FES), actually exercises the muscles," Dr. Hice said. "FES has been disregarded by many physicians and podiatrists, but some of our patients are apparently noticing an improvement in their use of muscles from the treatment."

While Steve receives treatment at PCPM, he will continue to perform and to live his life as fully as possible. Each new possibility for treatment offered by Dr. Hice and PCPM will take Steve's legs one step further toward strength and health.

Editor's Note:

Steve recently was a volunteer "model" patient for a class at PCPM. When faced with a captive audience, Steve lapsed into his performance character and literally broke up the class. Steve was favorably impressed by the considerate attitude of the students. In his words, "They treated me like a person."

Dr. Dyck

(continued from page 2)

pared to normal.

To prove our hypothesis we amputated legs of cats and evaluated nerve fibers above the amputations at various times after injury. Even by four months the frequency distribution peaks of myelin fiber diameters had shifted to smaller diameter categories. By one year this atrophic trend was clear. By two years the findings were very striking; an atrophic process had occurred. We could reproduce all of the changes that we had seen in uremic nerves. After injury the following sequential changes had taken place: axonal atrophy, myelin wrinkling, paranodal or internodal demyelination, remyelination, and with further atrophy axonal degeneration. Our model had proven what we had suspected from the human observations. Axonal atrophy may cause myelin remodeling.

A new insight which has come from our recent studies is the important role of environmental factors in the clinical manifestations of HMSN. This should not have been a surprise since it was already clear from both experimental study and human neuropathy that environmental factors do play a role in the expression of mutant genes. For example, in porphyria the clinical disorder is not expressed until you give the patient a barbiturate. In the case of Refsum's disease, a peroneal muscular atrophy with excessive storage of phytanic acid, deleting phytanic acid from the diet can improve nerve function. So in those two

genetic diseases, there is clear evidence that the environment does make a difference. It may also make a difference to the symptoms of HMSN neuropathy.

We have not talked about this yet, but physicians should make very careful distinctions between the symptoms which a patient has, the deficits which he has and the accompanying neurophysiological abnormalities. The physician should further make a distinction between conduction velocity on one hand and abnormalities of amplitude and distal latency on the other hand. They do tell you different things. Even though there is an association among them, they do represent different pathophysiological mechanisms, obviously related.

Let's deal with the issue of burning feet. When I first began to see patients with burning feet, it often occurred to me that perhaps they were neurotic patients, old age patients who had hurting feet and were complainers. But as I began to see these patients, I recognized that sometimes their symptoms were inherited. When I studied some of the older members of such a family, there was unequivocal evidence of neuropathy.

That was an important insight for me because here the deficit was not the problem, but the symptom was a major problem. A secretary from Phoenix, AZ came to see me because of burning feet without evidence of neuropathy. We did extensive autonomic and other tests and even nerve biopsy, all of which were normal. Her

(continued next page)

Dr. Dyck

(continued from page 5)

father, however, who had had burning feet, clearly had some signs of the electrophysiological abnormality of neuropathy, as did an uncle. The issue which was of such great interest to me was that environmental factors related to that symptom. For example, if the girl was in Minnesota, where it's cool and wonderful and placid, she did not have burning feet. In Arizona, where it's hot and they have only asphalt, she had a lot of pain. In other words, the thermal condition which her feet were exposed to affected her symptoms. Her uncle who plowed the fields found that he could get relief for his symptoms by taking off his shoes and walking in the cold furrow. In the winter time he would put his feet up against the wall to get relief.

So think carefully when you see a patient with these symptoms, because you might be able to ameliorate them. I have had patients coming from San Francisco who ran twenty miles a day. By reducing that to five miles a day they can live with their feet with their mild inherited neuropathy. So mechano-sensitivity is something you can modulate...it's an important issue, I think, that we should be thinking very carefully about modulation of neurophysiological activity which may give symptoms...

Sabin and Swift had a patient with leprosy, lepromatous leprosy. That is sensory loss in both hands. The man developed a stroke, so one arm was paralyzed. It was the other arm that developed the mutilating acropathy.

So here was an example of a patient who had the same degree of sensory loss bilaterally, but it was the use and abuse that led to the complications. So quite a different idea came to me about how people develop the mutilation. ...Not only is it the sensory loss, which may be trivial, but it's how you use your limbs, what precautions you take, whether you are indifferent to your injuries and whether you neglect trivial injuries. So you have an accumulation of trivial injuries which lead to these terrible complications. That is where all of us come in, because these are preventable troubles. Some of the major complications, the plantar ulcers, the Charcot joints are preventable. The patients who have inherited neuropathy should know that this is a failure of taking proper precautions.

In the Quebec and Virginia kinships that we studied, we found that it was the males that had the problem. The women looked after their feet, they did not walk in the frozen nights of Virginia and the crates of chickens did not fall on their feet. The outcome was strikingly different due to different environmental factors, but the sensory loss was the same. So a very important message to patients and to us: prevent these kinds of complications. The environment does influence the expression of these genetic abnormalities by what we do and what we advise. We wrote an article on the factors that may be involved...

I want to end up with what one tells the patient with inherited neuropathy. First of all, tell the truth. When I first began as a neurologist at Mayo Clinic I asked Clark Millikin, "What do I do?"

He said, "Tell the truth." That is not bad advice. I find that many physicians are kind of playing games with patients and there is no reason why you cannot tell the truth. The truth is generally better than the anticipation of an anxious patient. Second point: tell the patient about the disorder. Tell them what you know. When you do not know, say, "I don't know what you have." If you think the patient has inherited neuropathy, tell the patient what you need to do to be sure. A little candor can go a long way.

Tell them about the outlook. Some physicians follow the practice of sending the child out of the room while the disease is discussed with the parents. Don't do this! This practice sends the child a message that he must have something terrible that can't be talked about with him present. You can always talk about the outlook of a disease if you do it honestly. Many physicians paint much too gloomy a picture. They have been brainwashed

(continued on page 8)

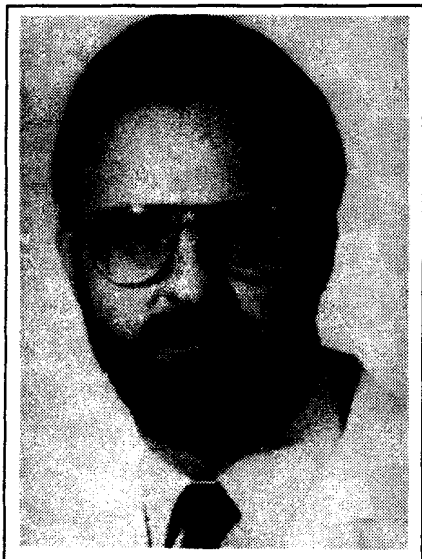
The **NFPMA Report** is published by the National Foundation for Peroneal Muscular Atrophy, a tax exempt not-for-profit corporation incorporated in the Commonwealth of Pennsylvania (established 1983).

This issue was designed by Mary Cawley Tracy and Lydia Brennan. (215) 878-4731 (215) 477-6206

Letters and inquiries may be addressed to:

Editorial Staff, **NFPMA Report**
National Foundation
for Peroneal Muscular Atrophy
University City Science Center
3624 Market Street
Philadelphia, PA 19104
(215) 664-6010

Ask the Doctor



Dr. Donal Costigan is an assistant professor of neurology at the Emory Clinic in Atlanta, GA. He is an active clinician and very interested in peripheral neuropathies. We welcome his response to the question posed by a CMT patient and thank him for his learned contribution.

Dear NFPMA:

In a recent issue of the NFPMA Report, Dr. Donal A. Costigan was quoted as warning of the potential harmful effects of certain drugs on CMT patients - among the drugs listed was Pyridoxine (Vitamin B₆). As a CMT patient, I am most interested in learning more about this drug and its effects. ...

After a bad bout with the flu ... my physician prescribed two tablets per day of All-B-And-C vitamins (containing B₆). Two months later I developed pains in the frontal muscles of my lower legs, which persisted for another two months. After seeing the NFPMA Report warning, I discontinued the All-B-And-C tablets ... and within two weeks the pains subsided. Of course, this may be entirely coinciden-

tal, especially since the B₆ content in the tablets was quite modest.

I would greatly appreciate any recommendations ... of some material on this subject or perhaps the address of Dr. Costigan.

E. J. S.

Brooklyn, NY

Dear E.J.S. :

Thank you for your letter which reached me via the NFPMA .

You report the development of possible additional symptoms of neuropathy after taking two tablets daily of ALL-B-AND-C, a multivitamin compound. The amount of Vitamin B₆ per tablet is 5 milligrams, making a total daily dosage of 10 milligrams.

It is safe to assume that people who suffer from hereditary motor sensory neuropathy, or CMT neuropathy, are more vulnerable to some substances which would be neurotoxic only in higher doses in the normal population. I am not aware of the "safe dose" of Vitamin B₆ in patients with HMSN/CMT but let me tell you what doses are considered hazardous for normal individuals. Schaumburg and colleagues (reference ¹) reported the development of a predominantly sensory neuropathy in people taking 2 grams or more of Vitamin B₆ daily, improving on discontinuing the treatment. This would be considered "mega-vitamin" therapy. In a subsequent report (reference ²) a woman became symptomatic on 500 mg. per day, again recovering when the drug

was discontinued.

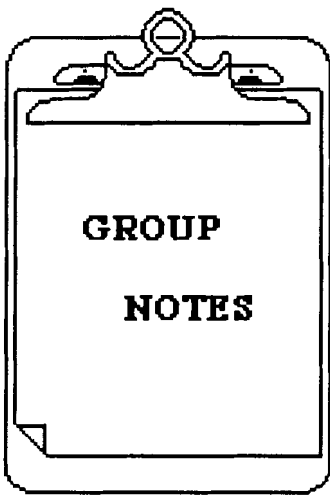
My advice to patients suffering from HMSN/CMT, and indeed any significant neuropathy, would be to avoid the use of Vitamin B₆ unless the prescribing physician has a strong suspicion that Vitamin B₆ is actually deficient. For example, there are situations in which administration of another drug may antagonize the effects of B₆, and it becomes necessary to give a compensating dose. However, most people absorb Vitamin B₆ sufficiently well that a balanced diet suffices, even while convalescing from an illness.

Of course, there is no way of proving a connection between your exposure to 10 mg. of Vitamin B₆ per day and the development of your symptoms. They may not have been due to neuropathy as you yourself point out. However, your letter is very instructive to HMSN/CMT neuropathy patients, and emphasizes the need for patient awareness. Many doctors may not necessarily know the broad range of common substances which can harm the peripheral nervous system. Regardless of your particular case, we will have to assume that unusually small quantities of Vitamin B₆ can cause damage in patients with pre-existing neuropathy.

Donal A. Costigan, M.D.

¹ Schaumburg, H., Kaplan, J., Windebank, A., et al. *Sensory Neuropathy from Pyridoxine Abuse: A New Mega-Vitamin Syndrome*. N. Engl. J. Med. 1983; 309:445-8.

² Berger, A., Schaumburg, H. *More on Neuropathy from Pyridoxine Abuse*. N. Engl. J. Med., 1984; 311: 986-7.



A primary goal of the NFPMA 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 NFPMA program, inform and support anyone who must deal with this often overlooked disease.

There are already several NFPMA support groups. These chapters are spirited and growing stronger, but more groups are needed in other parts of the United States. The NFPMA will gladly help you to set up a chapter in your area. For information contact the NFPMA by mail or call (215) 664-6010.

Perhaps there is a chapter meeting near you. You are cordially invited to join these groups in their upcoming events.

New York

Meeting: April 8, 1989
Where: Rusk Institute of Rehabilitation Medicine
Room RR 610 (6th Fl. Research Wing)
400 East 34th Street (at First Avenue)
New York, NY
Time: 1:00 - 4:00 PM (every other month)
Contact: Linda Phillips Goldfarb (212) 496-0001

New Jersey

Meeting: April 8, 1989
Where: Englewood Hospital
Clinic Conference Room
350 Engle Street
Englewood, NJ
Time: 10:00 AM (every other month)
Contact: Ann Lee Beyer (201) 391-4624

Delaware Valley

Meeting: April 8, 1989
Where: Holy Redeemer Hospital
Meadowbrook, PA
Time: 10:30 AM
Contact: Rex Morgan, Jr. (215) 672-4169

Tidewater, VA Area

Meeting: April 29, 1989
Contact: Mary Jane King (804) 591-0516
Ellen Morton (804) 851-7046
Where: Riverside Hospital
School of Professional Nursing
J. Clyde Morris Blvd.
Newport News, VA

Greater Atlanta

Meeting: March 18, 1989
Where: 2885 Brookcliff Lane
Marietta, GA
Time: 2:00 PM
Contact: Molly Howard (404) 253-5632
Sue Saye (404) 565-5950

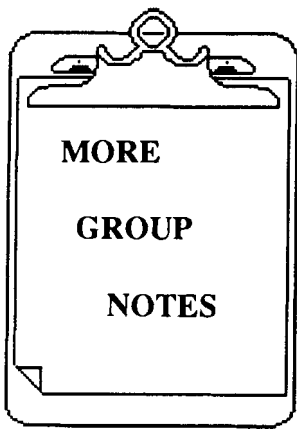
Cleveland, Ohio

Contact: Norma Markowitz (216) 247-8785

Orlando, Central Florida Area

Meeting: March 18, 1989
Where: 102 Hiawassee Road
Orlando, FL
Contact: Mary Beeler (407) 295-6215

(continued next page)



Fort Pierce Area, Florida
(Atlantic Coast)

Contact: Dorothy Stefanovich
(407) 461-1016

Greater Dallas, Texas Area

Contact: Karen Edelson, D.P.M.
(214) 542-0048

San Diego, California

Meeting: Monthly on the 3rd
Thursday, 6:00 PM

Contact: Gary Oelze
(619) 792-1427

San Francisco, California

Meeting: Spring '89

Contact: David Berger
(415) 461-3906
(after 6 PM)

Parsons, Kansas

Meeting: Spring '89

Contact: Tammy Taylor
(316) 421-5286

Where: Labette Community
College, Parsons, KS

Dr. Dyck

(continued from page 5)

by the textbooks. The textbooks tell the worst story. The truth is usually better than the textbooks by a long shot. You may have noticed that our slides, our textbook pictures showed only the worst cases, but 95% of cases have less abnormality than demonstrated in lectures and chapters.

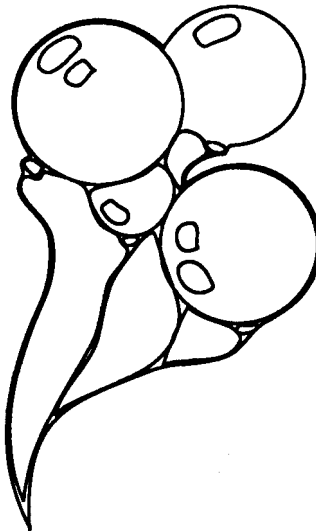
Tell them about the outlook. The outlook is excellent in most cases. They can go to school, they can marry, they can have children. Why not? Why should they not have children? Some of the best people I have known have Charcot-Marie-Tooth disease. Tell them about what they should do. In my experience,

they all want to be geologists or foresters. Well, try to persuade them to pursue other jobs. They should go to school and learn something else which also would be worthwhile, besides it might be fun.

Tell them about how their disease is inherited. Do not go simply by the phenotype; base it on the examination of the relatives. You may need to examine the parents, the children, the uncles, and the aunts. Get them to come in. Do not get someone in San Francisco to do it, do it yourself.

If you do not adequately advise about inheritance, a legal problem may ensue. I have known of cases where doctors have told patients that they had an inherited neuromuscular

(continued on page 10)



NEW GROUPS !

Rochester, NY

Meeting: May 7, 1989

Contact: Neale Bachmann (716) 554-6644
Bernice Roll (716) 584-3585

Boston, MA

Contact: Eunice Cohen (617) 894-9510

Indianapolis, IN

Contact: Robert Birdwell (317) 352-0235

Chicago, IL

Meeting: April 1, 1989 (1PM)

Where: Rush-Presbyterian-St. Luke's Medical Center (Rm. 539)

Contact: Stuart Feen (312) 680-7709

NFPMA Dedicated Contributions

In Memory of Irving J. Finkelman

Madelyn & Larry Lesnick	Samuel Rappaport	Marilyn & Martin Berger	Max & Robin Weinberg
Rose & Charles Bock	Terrie & Gene Rosengarden	Mark B. Frost, Esq.	Harold A. Boden
Sheila D. Davis	Martin Greenberg	Juliet C. Welker	Sandy & Marvin Melnikoff
Harriet, Franklyn, Steven & Sharon Weiss			

In Memory Of	Contributor	In Memory Of	Contributor
Milton Kunken	Miriam & Paul Moskowitz	Turissa W. Hawkins	Mary Jane & Bill King
Milton Kunken	Doris Shapiro	Robert G. Bradwick	R. Gordon Bradwick
Milton Kunken	Libbie Pitegoff	Evelyn Margaret Bell	David Bell
Milton Kunken	Mr. & Mrs. Sylvan Shapiro	Harley Bales	Geraldine Yount
Harry Brezel	Rose Edelson	Eleanor Lowy	R. C. Andrews
Harry Brezel	Dr. Karen B. Edelson	Jacob J. Masel	Edythe J. Masel
Sidney Bedell	Harry & Shirly Green	Norman Kunken	Mr. & Mrs. Sylvan Shapiro
Sidney Bedell	Mr. & Mrs. Sylvan Shapiro	James V. Ralston	Barbara H. Jaeger
Richard H. Hawkins	Jeanne Morton Hawkins	Doris Garboden	Mr. & Mrs. Sylvan Shapiro

In Honor of Hart & Minna Wurzburg

Priscilla S. Sloss	Mr. & Mrs. G. B. Frank	The Stines	Joell & Vernon Fox
Mr. & Mrs. L. Pfaelzer	Mr. & Mrs. A. J. Weiss	Mr. & Mrs. H. J. Levi	Lois Esserman
Mr. & Mrs. G. F. Baer	Helen & David Liebman	Mr. & Mrs. R. Wile	Mr. & Mrs. A. N. Seessel, Jr.
Mr. & Mrs. Robt. I. Logan	Marge & Bill Sherman	Mr. & Mrs. A. R. Wolff	Mr. & Mrs. E. Goodkind
Mr. & Mrs. Heorz Kahn	Mr. & Mrs. S. T. Lawton, Jr.	Mr. & Mrs. H. B. Solomon	Mrs. R. Oppenheimer
Mr. & Mrs. A. Adler, Jr.	Mr. & Mrs. L. Lepman, Jr.	Mrs. Albert Orlin	Mr. H. Sonnenschein
Mr. & Mrs. J. Goldsmith	The Ullmans	Mr. & Mrs. M. Minkin	Mr. & Mrs. R. Alschuler

Ann, Trip, Kathy & Jamie Wolbach

In Honor of Eileen V. Perrella

Patricia & Francis Monahan
Eileen K. Daly & Robert K. Kelly
Jack R. Mennis
Robert & Eileen Daly
Dr. Donald J. Perrella
Mary Jo & John Murphy

In Honor of the Pearl Family

Frances & Sidney Dolled
Frances Joselit
Frances R. Hurwitz
Gerald Mazur
Shelton & Gloria Wolf
Dr. Stanley Teitel
Sean M. O'Connell

(continued next page)

Dedicated Contributions (continued)

In Honor Of

Henry P. Eldredge
Dennis L. Parker
Kathleen Balsamo
Mrs. Sheri Sachs
Joan Decker
Mrs. Sophie Bradwick
Elsa Johnson
Rebecca Sand
Freida Gottesman
Mr. & Mrs. Edward Sachs
Dr. Donald J. Perrella
Mark Weiss
Kyle Trattler
Dr. Frank S. Dingwerth

Contributor

Rotary Club of E. Greenwich, RI
Patricia Parker
Katherine E. Schnelle
Elsa & Gyula Kovacs
Anita M. Decker
Laura & Timothy McCann
Byron & Lucy Johnson
Janet Sand Lawner
Mr. & Mrs. Sylvan Shapiro
Mr. & Mrs. Arnold R. Wolff
Dr. Robert Goldberg
Janeen Weiss
Ross Trattler
Sherry & Kent Brown

Dr. Dyck

(continued from page 8)

disease but have failed to provide enough information about the implications for the descendents. The physician could be perceived as having been negligent if insufficient information was provided. To be the genetic advisor the neurologist should be able to explain Mendelian inheritance and the probability of involvement among offspring. Remember that in dominantly inherited HMSN-Ia and -b and also in HMSN-II, although the probability is that 50% of offspring of a person with the disorder will be affected, a lesser percentage will have the severity of involvement that the parent has. On average the affected children are going to be less affected than the affected parent. That's good news.

Physical fitness. Although we are all going to do the definitive study and

Wally Bradley has just told me about an excellent design, we know that physical fitness is good for patients. have had HMSN patients who were the tennis champ of Indiana or the winning bicyclist of Iowa. They seem to have clearly helped themselves with exercise. Physical fitness also helps control weight.

Diet. There is no evidence that diet has anything to do with this disease. So, except for the amount of calories, there is no information.

Vitamins. Don't waste your money; it's been tried for fifty years. It never did any good. Unless you are B-12 deficient, are a lactating mother or were a prisoner of war, you do not need exogenous vitamins. Excessive B-6 vitamin can cause neuropathy!

Ultimately, the point I want to make is to provide hope. That is what being a

physician is about. When you talk to patients with this disorder, tell them that for most patients with HMSN the life expectancy is normal. Some of the life insurance companies do not seem to be aware of this fact! The fact is that life expectancy for most of these patients is normal. There are many worthwhile things to do. "Ask not what the country can do for you, but what you can do for the country." Most patients with HMSN should hold jobs. Many patients with HMSN are too concerned about their disease. There are some mighty good things they can do for others, and it also minimizes their concern about themselves.

Tell them you don't want to see them for ten years, no, twenty years. You see, what that tells them is that they are not on a precipice, they are not about to fall off. Yes, you will see them in a year, but get across the idea that there is nothing to be alarmed about. There is hope in that the people here are going to tell us where the CMT genes are, and are going to find the specific proteins, and are going to find the specific treatment for the disease. So there is considerable hope that the causes of HMSN will be delineated in the next few decades and good prevention and treatment will become available. Already there is a great deal that can be done symptomatically!



*If you move,
send us your change
of address.*

Enclose a former *NFPMA REPORT* mailing label.

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 returning this form. 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:

NFPMA Report

University City Science Center
3624 Market Street
Philadelphia, PA 19104

Name: _____

Address: _____

Telephone: (_____) _____

Tell us about yourself (optional):

- ☐ CMT patient ☐ Interested supporter
☐ Medical professional ☐ CMT family member
☐ Research scientist ☐ Other _____

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 are listed below. Single lecture tapes (1 hour, 15 min.) rent for \$10, and the double lecture tapes (2 1/2 hrs.) rent for \$15. The rental fee includes prepaid return postage. To order a tape, fill out our *Get in Touch* form and send it with a check or money order to the NFPMA.

Please check the appropriate boxes:

- ☐ Mailing List Request
☐ Tapes (Below)
☐ Physical Therapy - \$10 ☐ CMT Neurology - \$10
☐ Orthopedic Surgery - \$10 ☐ CMT Genetics - \$10
☐ Physical Therapy/Orthopedic Surgery (one tape) - \$15
☐ Neurology/Genetics (one tape) - \$15

NFPMA REMEMBRANCES

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 or anniversaries. They also make thoughtful thank you gifts. You can participate in the memorial and honorary gift program of the NFPMA by mailing the form below with your check to NFPMA, University City Science Center, 3624 Market Street, Philadelphia, PA 19104.

Name: _____

Address: _____

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: _____

Amount Enclosed: _____

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



F. Y. I. ...

**Back issues of the
NFPMA Report are available at
\$2.50 a copy.**

Volume 1, Nos. 1,2, and 3

Volume 2, Nos. 1,2, and 3

**Write or call the NFPMA.
(215) 664-6010**

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

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) 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.

THE NFPMA REPORT

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

University City Science Center
3624 Market Street
Philadelphia, PA 19104

**Bulk Rate
U.S. Postage Pd.
Ronkonkoma, NY
Permit No. 1426**

TO: