

THE CMTA REPORT

Spring 1993

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Providing information on Charcot-Marie-Tooth disease (a.k.a. Peroneal Muscular Atrophy or Hereditary Motor Sensory Neuropathy), the most common inherited neuropathy. Contents © 1993, CMTA. All rights reserved.

Taking Charge of Your Disease

by Ann Beyer

"It has taken years and thousands of dollars plus a great deal of time and effort to get diagnosed."

"There is such a lack of knowledge about CMT that the medical treatment I receive is often inadequate and at times even harmful."

"I am frustrated that there is so little interest in CMT and that so little research is being done."

"It is difficult, emotionally and physically, to live with a progressive, hereditary illness."

"I thought I was the only one in the world with CMT."

"There is the devastating reality that not much can be done to alleviate the physical symptoms of CMT."

For the past ten years, I have not only been dealing with Charcot-Marie-Tooth in my own family, but I have also been a volunteer with the CMTA - at both a local and national level. Because of my involvement, I have heard the above comments from many individuals who, like me, are affected by CMT.

However, I am not helpless, and you are not either! As a result of several conversations, especially one with Mary Beeler, who started the Orlando support group, I have put together a workshop called, "Taking Charge of Your Disease: How Not to Be a Victim."

The purpose of the workshop is to inform people that there is a great deal individuals can do about CMT and to encourage them to do it. I use the word "victim" in the title of the workshop because as long as we look to others to solve our problems, we are victims. But, there is no reason for any of us to put ourselves in the "victim" position. We cannot do anything about the disease itself; it is and



Ann Beyer

will always be part of our lives, but we can do something about how we let it affect us. This article is taken from the workshop and offers eight suggestions for taking charge of your disease.

1. Become knowledgeable about CMT. Learn all that you can about this disease, especially the type that affects your family. (Given the recent breakthroughs that have taken place, knowing what type you have is extremely important.)

The CMTA has several publications that will help you become better informed. Thoroughly read the newsletter. It is an excellent source of knowledge and provides advice on how to handle CMT, up-to-date information on medical advances and ongoing research, and information on upcoming conferences, workshops, and support group activities.

Order several copies of the association brochure on CMT. Familiarize yourself with it. In the brochure - as well as the newsletter - there is a list of neurotoxic drugs that are potentially harmful to people with CMT. It is important that you know what these medications are so that you can avoid taking them.

There is also a 16 page booklet, CMT FACTS I, which gives an excellent overview of CMT. (Editor's note: CMT Facts II will be available in June, 1993.)

Obtain and carry the CMTA medical identification card which is invaluable in case of accident or medical emergency. On the back of the card there is the list of the neurotoxic drugs. When you send for other materials, also ask for the ID card. If there are others in your family with CMT, ask for several.

2. Educate the health practitioners who treat you and your family members. This includes all of the health care providers that you see on a regular basis. Encourage them to join the CMTA and

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Encourage them to join the CMTA and read the newsletter. They treat you and members of your family; therefore, it is important that they know about CMT and especially the list of drugs which might be harmful to you.

When your physicians prescribe medications, remind them of this list and ask if the medicine is safe for you. As an added precaution, double check with your pharmacist before getting a prescription filled. These steps not only ensure you will receive better medical treatment, but they also make health care professionals more aware of CMT.

3. Sit down with your family and friends and talk to them about CMT. Tell them that you want them to know and understand what it is like to have the disease. Give them a brochure and ask that they read it. It will not only help you, because they will have a better understanding of how you are affected by this disease, but by talking to friends and relatives, you are making even more people aware of CMT.

4. Get into a regular physical therapy program. The physicians who specialize in CMT report that it is important for people with CMT to be involved in a physical therapy regimen. However, I find that many people I talk to are not aware of this. A regular exercise program can make a big difference in how you feel. Needless to say, it is necessary to find a physical therapist who is familiar with CMT or one who is willing to learn more about it. For those of you who go to an MDA clinic, the MDA will provide up to two visits per year. Do the exercises the therapist recommends; they can only benefit you.

5. Find people who will listen - individuals with whom you can share your problems, feelings, thoughts, fears and triumphs. One of the best places to do this is in a support group. Belonging to one also removes some of the isolation that comes from having a rare disease. If you are not already a member of a support group, get involved in one. I cannot emphasize this enough. If there is no support group in your area, start one; it is not difficult, and the CMTA will help you.

A support group, however, is not just a place for talking about problems. It is also a place for learning. Not only can you learn more about how to deal with CMT and the problems it presents, but also what kinds of exercise seem to work

best and how to find the physicians and therapists who know and understand this disease. Being an active member can really make a difference in how you handle your disease.

6. If you are having a difficult time coping with CMT, consider counseling. Physicians rarely recommend it, but counseling can ease some of the emotional pain that can come from having the disease. Living with a progressive, hereditary disease can be very trying. Sometimes it can seem overwhelming. Do not hesitate to seek help. In the same way you look to health care professionals to help you deal with physical problems, you owe it to yourself to take care of emotional needs as well.

The Muscular Dystrophy Clinics have therapists available, and if you cannot afford a private therapist, ask about their services. Here's a word of caution. Some therapists and physicians have a tendency to downplay CMT. It is not unusual for people with CMT to hear, "What's the problem? You can still walk." Do not accept this treatment from anyone. Your worries and fears are legitimate and you have a right to your feelings. Being able to walk does not mean you are not hurting.

If you decide to go to a therapist, find someone who is licensed. That is usually a social worker (ACSW or CSW) or a psychologist (Ph.D.). In many states there are no licensing requirements for therapists, and a degree or two does not necessarily mean that the therapist is trained in psychotherapy or counseling. If the therapist is not licensed, stay away.

7. Support the CMTA. It is member-supported and a non-profit organization. There is no government money coming into the CMTA. Except for one paid employee, all of the work of the CMTA is done voluntarily. The workers include the President Karol Hitt, the Board of Directors, and the Medical Advisory Board, chaired by Dr. Robert Lovelace, Professor of Neurology, Columbia University College of Physicians and Surgeons.

The CMTA is finally in a position where it can begin to take off and get this disease known, but it needs your help. This is a patient organization and it is up to us who are affected by this disease - to support it. Be generous and encourage family and friends to donate also.

8. Read some of the literature on the interaction of mind and body. Our minds and our bodies are connected and affect one another. Our thoughts and

emotions play an important role in how we handle a chronic illness. Several books that deal with this subject are:

- **The Healing Heart and The Anatomy of an Illness** by Norman Cousins.
- **Love, Medicine and Miracles** by Bernie Siegel.
- **Mending the Body, Mending the Mind** by Joan Borysenko.
- **Healing and the Mind** by Bill Moyers.

Take charge and don't be a "victim."

§



Referrals Available

The CMTA has compiled a list of neurologists, orthopedists, physiatrists (a physiatrist is a physician trained in physical medicine and rehabilitation) and podiatrists who have a special interest in CMT. We can also access respiratory specialists. Additionally, we have listings for pedorthists. A pedorthist is a practitioner who provides care to the patient by fitting orthopedic shoes and devices, at the direction of and in consultation with physicians.

To receive any of these referrals send a stamped self-addressed business-sized envelope indicating the geographic areas needed to: CMTA, 601 Upland Avenue, Upland, PA 19015.

For referrals for a hand surgeon contact the American Society for Hand Surgery, 3025 South Parker Road, Suite 3025, Aurora, CO 80014, 303/755-4588. §

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The opinions expressed in the newsletter are not necessarily those of the Charcot-Marie-Tooth Association. The 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 disorders, these briefs offer current medical opinion that the reader may use to aid and supplement a doctor's treatment.

ITEMS of INTEREST



1. Demos Publications has announced the publication of a guide to pregnancy and birth in the presence of disability. The book is entitled: Mother to Be: A Guide to Pregnancy and Birth for Women with Disabilities by Judith Rogers and Molleen Matsumura. It is available for \$ 24.95 in softcover and can be ordered by calling 1-800-532-8663.

2. The Region II Genetic Center at The Children's Medical Center, Dayton, OH, is looking for pictures of babies, children and adults with birth defects, genetic disorders, or chromosome abnormalities for use in creating a "reality base" for educational programs for both public and professional groups. The genetic educator has slides of conditions, but they are clinical in nature and do not show the personality of the people. Do you have pictures which show both the characteristics of CMT and the personality of the person? Slides can be made from negatives, photographs or other slides. The pictures will be used for educational purposes, not-for-profit activities. Please call Betsy McFarland at 513-226-8300 if you are interested in sharing.

3. Closing the Gap, which is published 6 times a year, reviews hardware and software products appropriate for handicapped and disabled persons, and explains in everyday language how this technology is being successfully implemented in education, rehabilitation, and vocational settings around the world. Annual subscription rates are \$ 26. The February/March issue is the annual Closing the Gap Resource Directory, a comprehensive listing of commercially available hardware and software products identified by Closing the Gap as appropriate for special education and rehabilitation. The Resource Directory is available for \$ 12.95 plus postage and handling. Call or write Closing the Gap, P.O. Box 68, Henderson, MN 56044. 1-612-248-3294.

4. The recent and rapid advances in genetics are starting to result in negative as well as positive outcomes. In many ways, genetic technology has surpassed

the ability of society to manage its revelations, and has resulted in many difficult ethical and financial decisions. Carriers of genes for genetic disorders could lose some of their health care coverage or find it harder to obtain a job.

People who learn they are carriers of genetic disease will have to consider whether their insurance will pay for their children's health care, gauge whether they will be able to keep that insurance if they change jobs, and decide if they will even be eligible for life or medical insurance even if no one in their family has the disease. Insurance companies and employers already are using genetic tests to deny health care coverage to healthy individuals who are at risk for developing a disease years later in life. Although insurers may not demand genetic tests since they are very expensive (and they don't want to cover the cost), some are already asking for the results if a client decides to have the tests on their own. In one recent four year study, 93 cases of "genetic discrimination" were identified at one medical center alone in which insurers and employers made decisions to limit or deny health care coverage based on the results of genetic tests. In a government survey of genetic counselors and nurses, fourteen percent had clients who found it difficult to get or keep health insurance because of genetic test results.

People will also have to think about how their carrier status might affect their employment. In one government survey, for example, nine percent of the employers said they consider dependents' potential expenses when deciding whether to hire someone. Employers have also cut off health care coverage for families who have a child with a genetic disease.

Some states have passed laws to prevent certain kinds of "genetic discrimination" by employers or by life and disability insurers. But self-insured employers, who pay for medical care for more than half the employees in the country, do not have to comply with state laws. These laws are very narrowly focused, and the Americans with Disabilities Act does not specify carriers of disease genes among the "disabled" that it protects from discrimination. Although the California legislature passed a bill two years ago placing an eight year moratorium on the use of genetic tests to determine eligibility for health insurance plans, and would make it illegal to use genetic information to disqualify people from group life and disability insurance plans, the bill was vetoed by the administration.

MEDICAL ALERT



*Certain Drugs
Toxic to the
Peripheral
Nervous System*

This is a list of neurotoxic drugs which could be harmful to the CMT patient.

Adriamycin
Alcohol
Amiodarone
Chloramphenicol
Cis-platinum
Dapsone
Diphenylhydantoin
(Dilantin)
Disulfiram (Antabuse)
Glutethimide (Doriden)
Gold
Hydralazine (Apresoline)
Isoniazid (INH)
Mega Dose of Vitamin A
Mega Dose of Vitamin D
Metronidazole (Flagyl)
Nitrofurantoin
(Furadantin, Macrochantin)
Nitrous Oxide
(chronic repeated inhalation)
Penicillin
(Large IV doses only)
Perhexiline (Pexid)
Pyridoxine (Vitamin B6)
Vincristine

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

There is now no federal protection, and probably will not be any, if health care consumers do not make their concerns known to state and federal politicians. With the current interest by politicians in health care reform, this and the health care priorities issue would appear to be important areas for consumer political action groups to consider. (Neuromuscular Diseases Newsletter, University of California, Davis)

5. Disability Workbook is designed to help build a thorough case when filing for Social Security disability benefits. It is available for \$ 18.90, including shipping, from Physician's Disability Services, Inc., at 1460 Gov. Ritchie Highway, Arnold, MD 21012, (301) 974-1111. §

Triumphant Survivors



(Editor's note: This article is excerpted from *INSIGHTS* with permission of the Spina Bifida Association. Their toll free number is 1-800-621-3141.)

Ann Kaiser-Stearns is a woman who deals in adversity. Actually, she revels in it - or rather the quiet strength humans can acquire when they have the determination to conquer their adversity.

The best-selling author has helped millions deal with life crises such as job loss, death of a child, divorce, chronic illness, or the birth of a disabled child.

It is what Dr. Stearns learned about the strength of the human spirit that moves and motivates her. Her research has led her to conclude that people who have experienced crises fit into three categories:

- the permanently broken (bitter people who never seem to crawl out of their emotional state)
- those who cope reasonably well (go through the grieving stages and other normal reactions to crises) and
- people she termed "triumphant survivors."

The characteristics that differentiate the triumphant survivors, she says are that they are wise, happy, caring, sensitive, useful to others and capable of living the best life within reach.

In the aftermath of loss, triumphant survivors manage to deal with pain in small segments, Stearns said. That lesson was learned by General Robby Riesner while

he was a prisoner of war in Vietnam for more than seven years. Then Colonel, Riesner was a ranking officer and the target of torture. He was subjected to almost a year of solitary confinement in total darkness.

"I learned to say to myself, 'You can take it one more minute - just 60 seconds,' the general told Stearns. Recovering alcoholics and drug addicts also learn to take life one day at a time.

One of Stearns' professors at Duke University impressed upon her his belief that it is important to see life as a story that is yet to be completed.

This philosophy she has found to be true in her own experience. "I was suicidal for about two years," she explained. "I look back and think that I would have never dreamed when I was 27 that I would become a mother at 39 and have another baby three years later," she said of her adopted daughters.

Stearns said that when the love of her life was lost, she would not have thought it possible that she would have the chance to help a million people as an author. "Things may be difficult right now - even terrible - but when we are hurting, it is so important to remember that there is more to the story." To get to be a triumphant survivor, Stearns said, it takes lots of times of discouragement. "We are people who struggle with anger, depression, and self pity, but the lesson is, it is possible to make a decision that this is not how I want to live. Be determined not to stay in the victim role."

While interviewing General Riesner, Stearns said that she expected to see someone who felt that his country owed him something. But, like other triumphant survivors, Riesner knew others had also suffered, and he went beyond an "I'm entitled" attitude. As he said, "I'm entitled to suffer for awhile, and feel sorry for myself, but eventually, to be happy, we must say we won't get stuck in this place."

During the interview over breakfast in a cockroach-infested hotel, Stearns looked down at her scorched breakfast and felt sorry for herself. The now-famous General commented, "Isn't this a lovely breakfast? Let's bow our heads and thank God." His perspective was that compared to the scraps of food he re-

ceived in the POW camp, the rubbery eggs and black bacon was a veritable banquet.

Mrs. Anne Morrow Lindberg was interviewed on the Dick Cavett television talk show. The host asked about surviving the great adversities in her life, including having an infant son kidnapped and murdered, and watching her husband, Charles, go from hero to "goat" because he took so long to back efforts to go to war against Hitler.

Cavett asked her if her grace and strength came from suffering. Mrs. Lindberg threw back her head and laughed. "Oh, my dear Mr. Cavett, it isn't the suffering that makes you strong; suffering breaks you down. What makes you strong - what builds you up - is the loving and caring we get from other people as we are suffering."

Triumphant survivors, according to Stearns, connect themselves to others: friends, family, support groups, church or synagogues. "We need to experience normalcy and you get that by getting the support you need."

Triumphant survivors also find ways to say good bye to loved ones or to their dreams. They bring closure to certain events or stages in their life. Stearns finally brought closure to a part of her own life. Ten years after her divorce, she sent pictures of her adopted six-month old daughter from India to her ex-husband. "I was finally free to wish him a happy life." The sweetest revenge is personal happiness.

Stearns, like others, has found personal growth in helping others experiencing a similar loss. One woman, who had a colostomy due to Crohn's disease, also had the loss of her marriage to deal with. This woman found strength, and a new life, through visiting hospitalized colostomy patients. "I really believe that over half of us save ourselves from our own pain through helping others." Stearns concluded. §

HONORARIA

In Honor Of	By
Kim and Joy Lamer	Judith Lamer
Drucilla Lake	Betty Lou Smith
Margaret DeStefano	Joseph DeStefano
Marjanne DeStefano-Hill	Joseph DeStefano
Mrs. Hart Wurzburg	Mr. & Mrs. H.B. Solomon

MEMORIALS

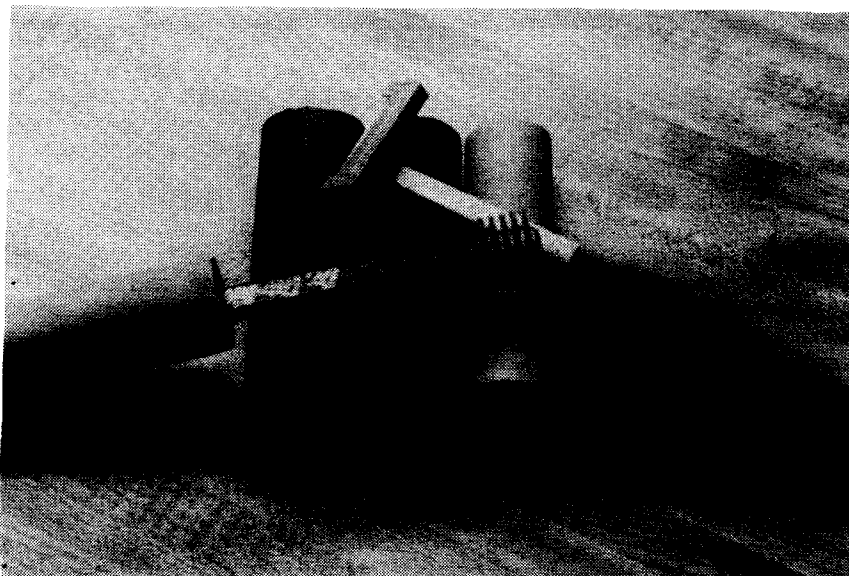
In Memory Of	By
Jarod Sizemore	Arita Sizemore
"Dop-POP" Bill Huntberger	Joy and Andy Dreibelbis
Kathryn Havill	Karol Hatt
Leo Breeden	Drucilla Lake
Ada May Faulkner	Marie Fortenberry

ADAPTIVE GADGETS: SUSAN'S TOP SIX

by Susan Salzberg, OTR



1. Amefa Vertical Knife is shown here with the angled fork and spoon which can be ordered with the angle for left or right hand use. These utensils are easy to grasp and are fairly light weight (spoon 1.5oz, knife 2 oz). The knife is especially wonderful as the grasp pattern allows for easier cutting. These are stainless steel and are dishwasher safe.



2. Cylindrical Foam: This is a very durable, closed-cell (doesn't absorb moisture) product that makes small handles larger. The 1/4" hole is good for a safety razor, toothbrush, pencil or pen and some forks and spoons. The larger holes are useful for hairbrushes, kitchen knives and tools.

Although I have read that "only" 75% of people with CMT have hand involvement, I think that we all do - some of us just don't know it yet! I am a forty-something female with CMT. Many years prior to my diagnosis, I chose Occupational Therapy (OT) as my life's work. I now find myself professionally advising people with weak hands. Most of them have arthritis. Many have neuropathies from diabetes and/or chronic alcoholism. Several of my patients even have CMT.

In medical practice the ultimate goal is to cure the patient. Your doctor can cure your sore throat and your stomach ulcers, but so far he or she can only "follow" your CMT. If you have specific complaints, your doctor may refer you to a rehabilitation professional - an OT, PT, speech therapist or respiratory therapist. Your quality of life may be improved by advice from a rehab professional.

Occupational therapists focus on a person's ability to do his/her "activities of daily living" (ADL's in rehab jargon). ADL's can be anything from how a person dresses, grooms and feeds him/herself to more complex tasks like cooking, caring for a child and/or adjusting to a work space. Since CMT is both a sensory and motor neuropathy, OTs look for both diminished strength and problems with coordination. My patients with weak hands benefit from my knowledge of various "gadgets" which help them do tasks that would otherwise be difficult or impossible.

The following are the Top Six Gadgets (by my personal vote) for people with weak hands. These devices make your grasp more efficient by either enlarging the grasp area so you don't have to squeeze as hard (#'s 1 & 2), changing the grasp pattern to a more efficient one (#'s 1 & 6) or by changing the method you use to accomplish the task (#'s 3, 4 & 5).

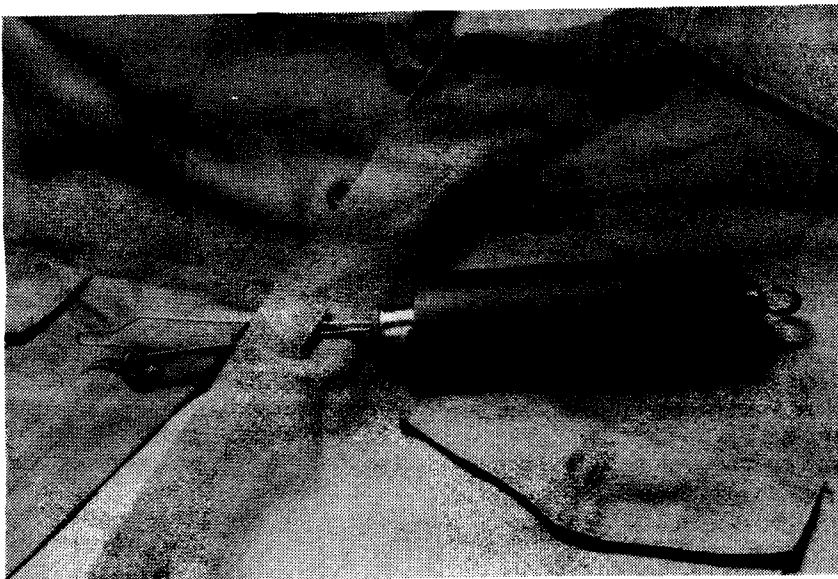
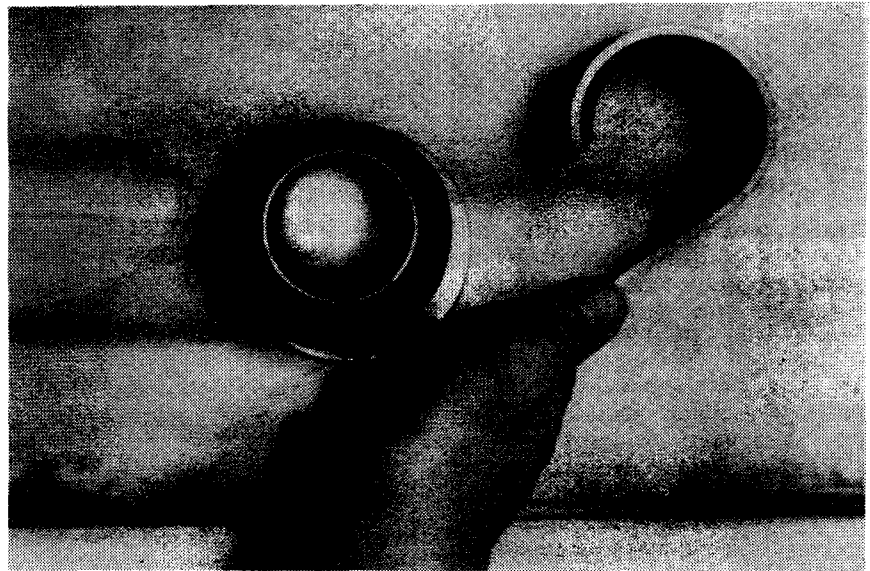
In the interest of space, only six gadgets are included here. If you order catalogs, you will see that there are hundreds of items to choose from, including devices to help with writing, cooking, bathing and dressing. Not every one is useful for every patient. Some, such as the button hook, come in several different styles which meet different grasp abilities. It is difficult to tell from a photo whether or not an item will be useful to you. As an OT, I recommend trying out any gadget before you purchase it. Most OT departments have many of these items available for patients to try out. Aside from recommending gadgets, an OT can instruct you in work simplification and energy con-

(concluded on back page)



3. Un-Skru Jar Opener: This must be mounted under the counter. I have a pretty respectable 40-45 lb. grasp, but find this device very handy - especially for new jars and soda bottles. You hold the jar with both hands and turn it using the large muscles of both forearms. The opener does all the gripping. I have been using mine for about 10 years and have given many as gifts to "normal" elderly folks.

4. Portable Door Knob Opener: This turns your door knob into a lever. It permits you to open a door by pushing with your arm rather than grasping and turning with your wrist. There are several varieties of door knob extensions, but this is the only portable one.



5. Button Hook: Long before my time, when women wore button-up shoes, they fastened them with a hook. I have been told that this gadget is a direct descendent. It comes in a variety of styles - wooden handle, foam handle, with or without zip hook. This one has a built-up handle with a zip hook at the other end. You stick the wire through the button hole, lasso the button, pull the button through the hole and then remove the wire from around the button. A needle threader works in much the same way. The hook at the opposite end allows you to zip your trousers without relying on pinch.

Letters to the Editor



Dear CMTA:

I am a CMTA member and a patient. I would like to request that you begin sending literature to my granddaughter as she also has CMT. My son is already on your mailing list.

I am purchasing all of the VCR tapes and I plan to show them to the doctors at two clinics. They seem to have missed the classes that covered CMT in medical school. It seems it's up to the patients to educate the doctors. I hope to interest some of them in joining you and receiving your newsletter and supporting your group.

Thank you for being there and speaking out and up! GOD knows you can feel kind of like a loner with this kind of problem. Your letter keeps me up-to-date and makes me see that I am not alone.

Keep up the good work and GOD Bless.

F.M. Wauzeka, WI

Dear CMTA:

I live in a small rural area where we have problems getting doctors to come to our area. Then, when they fill their obligations, they move on. Therefore, I don't have a doctor who knows anything about CMT and hasn't taken the trouble to find out. The doctor who diagnosed my CMT lives in Pueblo which is 120 miles away. It is hard for me to make the trip as I'm 81 years old.

But, thank goodness, I'm progressing very slowly and am still able to take care of myself.

I enjoy reading the CMTA Report. All I know about CMT I have read in your paper. Thanks so much.

G.S. Lamar, CO

**We want to hear from
YOU!**

Write us at:
Letters / The CMTA
601 Upland Avenue
Upland, PA 19015
215/499-7486



Dear CMTA:

Enclosed is my check, but also my many thanks for sending me copies of the CMTA Report.

As a patient with CMT, I find the report informative, plus a beam of sunshine regarding research.

The "Letters" section is always interesting to read. In the Fall 1992 issue, K.V. inspired me to check with my local shoe store about straps being added to pumps. What a help. Usually my visits to the shoe store were a "grand adventure." I thank K.V.

Please continue to send the Report!

J.W. Kearney, NJ

Dear CMTA:

Thank you so much for the Newsletter. I do appreciate it so much. I love to keep in touch with CMT. My husband had it. He is gone now. His sister and one brother have it now. I have grandchildren and great grandchildren and some of them could have it. No one knows where it came from. No one on either side of the family knows of anyone before them having it.

When my husband found he had it, no doctor knew anything about it so there was nothing done for him. He was 64 years old when the doctor found it. He lived to be 82. It was real hard on him. He had one hand and was a heart patient. He also had cancer, so many things were wrong with him.

I am in touch with the children's hospital in Ohio. Thanks again for your help.

M.C. Star City, AR

Dear CMTA:

I would like to thank you for publishing the letter from S.T. from Kansas City, MO. My son Jason, age 15, has CMT and has been experiencing some difficulties with his handwriting. I was not aware that medical coverage was available for the cost of a computer. I was also impressed to learn about the software program KEYWHIZ. I thank you for providing information that will help me prepare for the future.

Jason, age 15, has CMT with vocal cord paralysis. His CMT appeared spontaneously, with no family history of CMT. It

has not been determined to date if it is Type I, but I have been told it probably is not. Vocal cord paralysis is extremely rare with CMT. I am looking for anyone who has CMT with vocal cord paralysis. I would like to know their experiences to date and what problems they may or may not have encountered. I am also interested in any information that is available on vocal cord paralysis. The doctors do not seem to have any information and because of confidentiality will not provide me with the names of other parents or people with this problem. Please contact me evenings at (716) 398-2645 or write: Barbara Morse, 5677 Running Brook Rd., Farmington, NY 14425.

B.M. Farmington, NY

Dear CMTA:

How comforting to find out there exists an organization involved with informing and assisting patients and families, as well as medical professionals, in relation to Charcot-Marie-Tooth disorders.

It is only within the past year that my husband was diagnosed with CMTD. He had varied discomfort in his legs for several years, which we dismissed to the fact that he stands on concrete for an eight hour shift daily at his job. The pain and weakness seemed to be getting worse, and he sought medical explanations and treatment. His father had ankle, foot, and leg problems for many years, but we were not aware, until recently, that it was an hereditary condition.

An orthopedic surgeon could not diagnose the problem and recommended a neurologist. My husband saw a neurologist and had some testing done. After hearing that his father has CMTD, the doctor said that is my husband's problem also. My concerns at this time are to obtain a second opinion, to rule out any other possible problems or causes, and to find out what can be done to help him.

Please send me your physician's referral list, a copy of the letter to the Medical Professionals regarding the drug list, and the booklet, CMT Facts. Your information and your assistance is greatly appreciated.

K.S. Hagerstown, MD



Prenatal Diagnosis and Screening: What is it?



by Karen Copeland, M.S.

(Editor's note: With the availability of the chromosome 17 blood test to identify CMT prenatally, it becomes essential for young CMT families to understand what prenatal testing is and why it might be undertaken.) This article is taken from the publication TEXGENE, a newsletter of the Texas Genetic Network.

Pregnancy is a time of joy, anticipation and occasionally, anxiety. It is normal for couples to wonder if their baby will be normal and healthy. Most infants are born without problems, but birth defects do occur. Due to advances in prenatal diagnosis and pregnancy screening, it is now possible for couples to obtain information regarding certain aspects of their babies' health prior to birth; however, not all couples will benefit from prenatal screening and diagnosis. The decision to pursue testing is very individual. Listed below are some of the tools available to determine genetic risk.

BACKGROUND

Genetics is the study of the hereditary and environmental factors which affect our mental or physical development. There are approximately 100,000 genes (the smallest unit of heredity) present in every cell of our body, which determine all of our physical traits. Genes are packaged into structures called chromosomes. Congenital disorders (problems present at birth) and genetic disorders (inherited condition) affect two to four percent of all babies born. Problems can be inherited from the child's parents, caused by environmental factors, or there may not be a known cause. It is important to remember that there is a risk for birth defects in every pregnancy. The natural risk of two to four percent is present, even if there is no family history of problems, no pregnancy exposures or complications, and the couple is in good health. No medical test can eliminate or significantly reduce this risk. Genetic counseling can help determine if a couple's risk for birth defects is above that of the general population. Genetic counselors are health professionals with specialized training in medical genetics. They help families understand their risks and the testing options available to them.

FAMILY HISTORY AND OTHER RISKS TO THE PREGNANCY

A pregnant woman should complete a genetic screening questionnaire, which provides information to her obstetrician or midwife regarding: her health and the health of the baby's father; both family histories; previous pregnancy history; and, complications or exposures in the current pregnancy. She should discuss any findings, or areas of concern, with her doctor. Most questions can be addressed by the doctor, but occasionally, the woman may be referred for genetic counseling to determine if there is any increased risk.

BLOOD CHROMOSOME ANALYSIS (KARYOTYPING)

It is possible to analyze a person's chromosomes using a blood sample. This is only recommended if a couple has had: three or more miscarriages; a family history of mental retardation or congenital abnormalities; or, a previous child with a suspected chromosome problem. Most babies with genetic problems are born to healthy parents with normal chromosomes, so this test does not eliminate genetic risk or the need to consider other testing.

MATERNAL SERUM AFP (alpha-fetoprotein) AND OTHER BLOOD SCREENING TESTS

The serum AFP assay is a screening test for two groups of birth defects, neural tube defects (spina bifida and anencephaly), and Down syndrome. This blood test has been available for five to six years to all women; however, the test is specifically targeted to those women under age 35. AFP is measured in the mother's blood at 16-18 weeks gestation and the results are interpreted as either normal, high, or low. A high AFP value indicates an increased risk for spina bifida or other fetal defects; a low AFP value indicates an increased risk for Down syndrome. More recently, the test has been modified so that two other chemicals are measured in the mother's blood, beta HCG (human chorionic gonadotrophin) and estriol. Available for

only one year, this newer test goes by a variety of names. The combination of AFP, beta HCG and estriol increases the detection of babies with Down syndrome to 60-70 percent. The AFP portion of the test continues to detect 80% of babies affected with neural tube defects. When the test result is abnormal, additional testing must be performed to determine the cause. Causes other than birth defects include a twin pregnancy, incorrect gestational age, or a miscarriage. If an abnormal result cannot be explained, an amniocentesis is offered to clarify the result. Most women who have abnormal blood test results have normal babies. It is important to remember that the blood tests are only screening tests. They do not diagnose or eliminate the risk of a problem; they only increase or decrease the relative risk in the pregnancy.

ULTRASOUND

Ultrasound is a method of visualizing the baby through high-frequency sound waves, and does not have any known risk to the baby. Ultrasound examinations performed in the OB office are usually considered Level I. This is used to determine the gestational age, placental position and number of fetuses. A specialized ultrasound, known as high resolution or Level II, can look for structural malformations in the baby. It can determine that structures such as the limbs, heart, kidneys, brain and other internal organs are present and formed normally. It cannot detect minor birth defects, mental retardation, chromosome problems (such as Down syndrome), or other genetic diseases. An ultrasound, performed at 18-20 weeks gestation, is very useful in detecting some birth defects and provides many parents some reassurance. If problems are seen on an ultrasound, genetic counseling is usually necessary to discuss the findings and the possibility of additional testing.

AMNIOCENTESIS

Amniocentesis has been available for over 15 years and is considered a safe and accurate test. At approximately 15-18 weeks, an ultrasound is performed to locate a pocket of amniotic fluid. A thin needle is inserted through the woman's abdomen and uterus, into the fluid pocket, and a small amount of fluid is removed. After the cells in the amniotic

fluid are grown for one to two weeks, the chromosomes inside the cells can be analyzed to detect problems such as Down syndrome. AFP is measured in the fluid to detect neural tube defects. If a couple is known to be at risk for a diagnosable genetic disease (such as hemophilia, muscular dystrophy, sickle cell anemia, or cystic fibrosis), then specialized biochemical or DNA testing can be performed on the amniotic fluid cells. (Editor's note: Prenatal testing for CMT 1A is now available.) The risk of miscarriage due to amniocentesis is approximately 1/2 percent (1 out of 200) above the normal miscarriage rate. Amniocentesis is considered to be greater than 99 percent accurate in detecting neural tube defects. Occasionally, additional testing may be necessary to clarify an amniocentesis result.

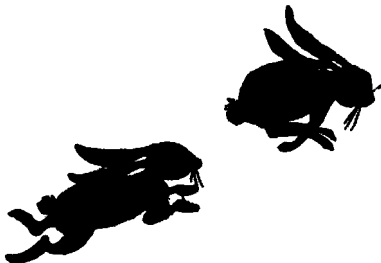
In recent years, amniocentesis has been performed earlier in pregnancy. The most common gestational age for "early" amniocentesis is 13-14 weeks, but some centers perform the test under 13 weeks. There is limited information regarding the safety of "early" amniocentesis. Some studies have shown the risk of miscarriage to be similar to standard amniocentesis; other studies have indicated a higher risk of miscarriage. There is no evidence of any other risks with the earlier procedure, but there is limited, long-term follow-up of the infants.

CHORION VILLUS SAMPLING (CVS)

CVS is a newer method of prenatal diagnosis and has been available for approximately six years. At nine to twelve weeks gestation, a small amount of placental tissue can be obtained using either a catheter placed through the cervix or a needle inserted through the abdomen. Ultrasound is performed before and dur-

ing the procedure to determine the best CVS technique. The placental tissue is grown and analyzed similar to amniocentesis. The accuracy of detecting chromosome abnormalities through CVS is greater than 99 percent. Women sometimes need additional testing to clarify a CVS result. Neural-tube defects cannot be detected through CVS; therefore, women having CVS are recommended to have a serum AFP at 16-18 weeks. The risk of miscarriage after CVS is estimated to be one-half to one percent higher than amniocentesis.

The prenatal screening and diagnostic tests described above are a method of providing information to parents about their baby. Most parents receive reassuring news, but some learn the baby has a birth defect. Currently, most of the problems detected through these tests are not correctable. Some parents may elect to continue the pregnancy and use the information for preparation and medical management. Other families may choose to stop the pregnancy because of the problems detected. This is a very emotional and difficult decision. Not all parents want to have this information. It is important to remember that choosing a particular test does not commit a couple to any other test or course of action. Parents need to be aware of the implications of prenatal diagnosis before having the test. Each test has advantages and disadvan-



Pharmaceutical Company Offers CMT Test

Genica Pharmaceutical Corporation, in collaboration with Baylor College of Medicine, announces the availability of a DNA based blood test for CMT type 1A. The test will be available in August, 1993. This test is based on the findings of Drs. James Lupski, Pragna Patel, and colleagues. Their work identified a DNA duplication on chromosome 17. This duplication has been found to be the cause of most cases of CMT type 1A. Contained within this duplicated region is the peripheral myelin protein gene, PMP 22, whose overexpression is now considered to be responsible for the disease.

The Winter 1993 issue of The CMTA REPORT, featured an in-depth "Research Update", reported by Dr. Kenneth Fischbeck, University of PA., which detailed the clinical significance of the PMP 22 gene discovery. It appears that the duplication of the PMP 22 gene accounts for the majority of both inherited and sporadic cases of CMT 1A. Based on finding that the chromosome 17 duplication is the common mechanism for CMT 1A, it has been suggested that this DNA based blood test may be the most suitable first test in the diagnosis of suspected CMT patients.

Technically, Genica's test will include an initial screening by Southern blot gene dosage analysis, followed by pulsed field gel electrophoresis (PFGE). The Southern blot assay will be informative for approximately 60-70 % of the population and will be reported to the physician within 2-3 weeks. For confirmations or mutations that require the PFGE technology (approximately 30-40 % of the cases) an additional time period will be necessary to confirm diagnosis of CMT 1A.

Genica's test, available in August, will be suitable for affected individuals and as yet undiagnosed cases. The test involves drawing two 10ml tubes of blood in yellow top tubes (ACD, acid/citrate/dextrose) and is shipped at room temperature. Genica's prenatal test will be announced when available.

Genica's technical service representatives can assist you in providing additional information, educational literature and sample procurement requirements for the test. Genica can be contacted at (800)-394-4493, and is located in the Worcester Biotech Park, Worcester, MA. §

CMT FACTS II Available in June

We are delighted to announce that after June 1, 1993, our second handbook of pertinent CMT topics will be available. CMT FACTS II is the sequel to CMT FACTS I, a publication that has been very well received. The new booklet is comprised of articles from past newsletters as well as new articles. Some of the topics covered are physical rehabilitation, vocational rehabilitation, orthotics, the Americans with Disabilities Act, children with disabilities, shoe suppliers, coping with a chronic disorder, as well as five pages of questions and answers from "Ask the Doctor." The neurotoxic drug list is also included with an explanation of each drug's probable use. CMT FACTS II is not a replacement for CMT FACTS I, but rather a second valuable addition to your CMT information library. The order form for this and other items available from the CMTA is located on the last page of the newsletter.

Support Group Notes

A primary goal of the CMTA is to become a truly successful advocate for those with CMT. Its message must reach the patients, their families, and the medical and research communities. Patient family support groups help carry out this function.

There are many CMTA support groups, but more groups are needed. The CMTA will help you set up a group in your area. For information about forming a group or being a local contact person please inform the CMTA by mail or call 215-499-7486.

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

Alabama - Greater Tennessee Valley
Bill Porter 205-386-6579W;205-767-4181
Meets at ECM Hospital, Florence, AL

California-- Los Angeles Area
Oxnard-Thousand Oaks
Janice Hagadorn (805) 985-7332

Adelanta (High Desert)
Mary'L Michels (619) 246-7807

Canyon Country - Saugus
Sheila Levitch (805) 254-5322
Denise Miller (805) 251-44537

California-- San Diego
Gary Oleze (619) 944-0550

California-- San Francisco
David Berger (415)491-4801

California-Santa Rosa
Freda K. Brown (707)573-0181

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

Florida- South
Robyn Cohen (407) 622-5829

Massachusetts-- Boston,
Donald Hay (617) 444-1627

Massachusetts- Southboro
Jim Lawrence (contact person)
(508) 460-6928

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

Michigan-- Detroit
Suzanne Tarpinian (313) 883-1123

Mississippi- Jackson
Julia Prevost (601) 885-6482
Henry & Brenda Herren (601) 885-6503

Missouri-- Kansas City
Sandra Toland (816) 756-2020

New Jersey-- Central
Janet Saleh (908) 281-6289
Somerset Medical Center
Sommerville, NJ 08876

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

New Jersey--Millville Area
Linda Muhlrig (contact person)
(609)327-4392

New York - Brooklyn
Alan Latman (contact person)
(800) 227-1343

New York City
Diana Eline (201) 861-0425

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

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

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

North Carolina--Eastern
Susan Salzberg (919) 967-3118
(919)286-0411 (x6586) days
Durham VA Medical Center

Ohio-- Cleveland
Norma Markowitz (216) 247-8785

Pennsylvania-- Delaware Valley
Dennis Devlin (215) 269-2600 work
(215) 566-1882 home

Pennsylvania--Duryea
Patricia Zelenowski (contact person)
(717) 457-7067

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

Utah-- Salt Lake City
Marlene Russell (801) 966-7563 home
(801) 565-1212 work

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

Virginia--Richmond Area
Dennis Breckenmaker (814) 748-9021
Steve Firestone (804) 745-4123

West Virginia- Central
Joan Plant (304) 636-7152 (after 6pm)

Washington, DC - Baltimore, MD
Lorraine Middleton (202) 362-4617

support group corner



Capitol Idea!

A new support group is forming in the Washington,DC-Baltimore,MD area. The group will hold its first meeting sometime in July (not yet announced) at the Kensington facility of Kaiser Permanente. The group leader will be Lorraine Middleton, 1-202-362-4617. Please call Lorraine and offer your assistance. We have waited a long time for a group to get going in this huge metropolitan area.

Mississippi



A new support group is forming in the Jackson,MS area. The group is under the leadership of Julia Prevost and Henry and Brenda Herren. The first meeting of the group will be on June 5,1993, at the University of Mississippi Medical Center, School of Nursing, 2500 North State Street, Jackson, MS. The speaker will be Dr.Luther Fisher, orthopedic surgeon at the University Hospital. Call Julia at 601-885-6482 or Brenda at 601-885-6503.

Last minute addition

CMT Conference
Colorado Springs, CO
September 18, 1993

Contact:
Dr. Gregory Stilwell (719)594-9920

Further details were unavailable at press time. More information will be provided in the Summer issue of *The CMTA Report*.



Ask the Doctor

Dear Doctor:

I have been having a problem with osteoporosis and my doctor feels that the problem might be related to my CMT. He wants to start me on estrogens and I am wondering if they will cause me any difficulties because of CMT.

The Doctor Replies:

The patient wished to know if there were any contraindications to taking estrogens or progesterone as a first line defense, and I have not encountered any difficulties with CMT patients who have been taking this hormone therapy. Neither do I believe that there is any theoretical reason why this should make a neuropathy worse. I believe that if there is any deterioration in the Charcot-Marie-Tooth disorder, she should be evaluated immediately, but I would not expect such, and believe she should take the advice of her gynecologist.

Dear Doctor:

I have been prescribed the drug Noroxin for a recurring bladder infection. I am interested in finding out if there would be any problem with my CMT that might occur from taking this drug.

The Doctor Replies:

The drug quinolone or Noroxin has been prescribed for a bladder infection, and the only problems of drugs prescribed for bladder infections is that one has to be sure that there is no problem with kidney or renal excretion in which high levels

may be inadvertently built up in the blood. Firstly, Noroxin is of correct pharmacopeial name norfloxacin or norfloxacin which is a quinolone derivative. The absolute contraindications are that it should not be used in children or pregnant women. As regards neurological side effects, none of these effect the peripheral nervous system which of course is the area involved in CMT, but the PDR (Physician's Desk Reference) does indicate that there may be some adverse reactions in the central nervous system and they have listed nausea at 2.8%, headache at 2.7% and dizziness at 1.8%. Should the patient develop these, she should consult her physician and should there be any worsening in her CMT, we should try to obtain objective evidence of this in case this is a drug that should be put on the proscribed list.

Dear Doctor:

I was diagnosed with mild CMT in 1992. Just recently in January of 1993, I had a dorsal column stimulator attached to my spine with a receiver under my arm towards the back. Even though this was not considered major surgery, I have had serious after effects which I can only assume are due to my CMT. I can hardly walk as I tire very easily now. I have days when I am very weak and exhausted. The DCS was done because of severe pain, nerve damage, and scar tissue from 5 previous back surgeries.

My question is, will I recoup what I have lost with regard to strength? If I have any

other surgeries will my CMT get worse each time? I am 49 years old and not dealing well with this present situation. Is pain involved with CMT? If so, could mine be amplified due to my back surgeries? What effects will pain medication have on my CMT?

The Doctor Replies:

When a person has back pain requiring 5 previous back operations for relief of pain, nerve damage, and scar tissue, I would consider any further operative intervention, especially that of putting in a dorsal column stimulator to be a major undertaking. It also represents a further mode of treatment.

Without any significant clinical information, I would feel that if there is any significant loss of strength quite rapidly, it would not be caused by the slowly progressive nature of Charcot-Marie-Tooth disease. The immobilization, change in activity, continued pain, possible weight gain or weight loss takes a long time to recover. Although Charcot-Marie-Tooth disease does affect the spine, especially in the teenager and young adult, presenting with kyphosis, scoliosis or other types of spine deformity, unless a severe fixed curve is present, this is not generally associated with pain.

The use of pain medication does affect CMT patients because it diminishes their alertness, causes muscle relaxation and in many instances leads to decreased activity as well as possible other side effects.

Pulmonary Function and Restrictive Lung Disease Profile

(Editor's Note: This article is one segment of a research update on the Impairment and Disability Profiles of Neuromuscular Diseases: Hereditary Motor Sensory Neuropathy (CMT) as published by the Department of Physical Medicine and Rehabilitation at the University of California, Davis.)

Pulmonary function tests (PFT) were obtained in a sample of 40 individuals from the clinic population. There was only mild impairment on routine (spirometric) PFTs with the group mean for most parameters within normal reference standard limits. Vital capacity was less than 80% of the predicted (80% or above is considered normal) in 33% of the individuals and less than 50% of the predicted (50% or less is considered to be severe involvement) in only 8%.

There was greater impairment, however, in the static airway pressures. Thirty two percent of the individuals had abnormally reduced maximum inspiratory pressures (MIP) with a low of 35% predicted, and 76% had abnormally decreased maximum expiratory pressures (MEP) with a low of 29% predicted. MEP reflects abdominal and external intercostal muscle strength and MIP is primarily generated by the diaphragm. The results suggest that the expiratory muscles are more affected than the inspiratory muscles in HMSN, resulting in relatively normal spirometric PFTs since these tests primarily reflect active inspiratory function. There was no significant age or disease duration effect on PFT measurements with either one-time event or individual longitudinal analysis. The presence of spine deformity did not significantly affect the pulmonary function parameters. Although phrenic nerve (nerve to the dia-



(continued on page 15)

Siblings Also Suffer When a Child Isn't Well

by Barbara Meltz

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"Sometimes I wish she wasn't my sister," 12 year-old Todd Roy of Lynn, MA, says of Ryann, who is 9.

A typical enough comment, one that could be spoken by almost any sibling in America. But Todd's comment has an added level of poignancy because Ryann has a severe case of juvenile rheumatoid arthritis.

When a child has a chronic illness, whether JRA or asthma or leukemia, the most immediate impact, of course, is on the child and parents. But illness or disability also affects siblings. They can come to feel neglected and uncared-for, leaving them ripe for jealous, guilty, even angry and hostile feelings they don't understand.

Unfortunately, siblings are often the last ones parents, even professionals, pay attention to. "They tend to be the forgotten kids," says child psychiatrist David DeMaso, chief of emergency psychiatric services at Children's Hospital in Boston. He works with families of children with chronic illnesses.

It's not a given that siblings will have adjustment problems. But it is a given that these kids have parents who are sometimes unavailable to them or whose stress level is very high. While parents may be unable to change those facts, they can be open and honest with their children, says DeMaso. And that can go a long way.

"These siblings need to feel they are part of the family, part of the process," says pediatrician James Perrin.

Since Todd's sister was diagnosed two years ago, his mother Lisa, frequently talks to him about why Ryann needs so much help in the mornings or why she can't get out of the car by herself.

"He needs reminders, 'cause it looks to him like she gets special treatment," Lisa says. In some ways, it's easier to deal with her younger daughter. Renae, at 3 1/2, doesn't understand why "Sissy is so fragile," but she knows not to jump on Ryann or grab her leg or arm.

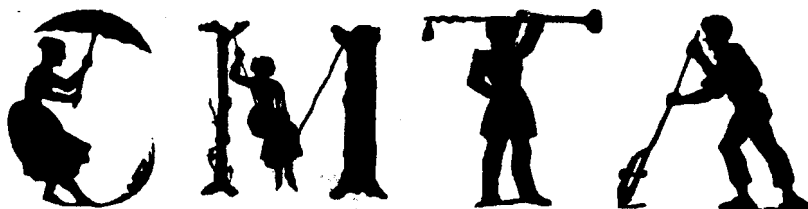
(continued on page 14)

Conferences, Conferences, Conferences

Three CMT patient/family conferences are scheduled for the Fall of 1993. The first of these conferences will be October 2, 1993 in Houston, TX, at Baylor College of Medicine. The host at that conference is Dr. Stanley Appel, chairman of the Department of Neurology. Among the speakers will be Dr. James Killian, research neurologist and clinician and Dr. James Lupski, noted CMT researcher. The second conference will be October 30 at the Crystal Clinic in Akron, OH. The conference organizer is Dr. Ian Alexander, noted orthopedic surgeon. He is assembling area CMT specialists for this all-day conference. The third conference will be on November 5, 1993, at Children's Hospital in San Diego, CA. Details of the California conference will appear in the Fall newsletter.

Each conference will be a full day of information about CMT from neurologists, orthopedic surgeons, physical therapists, geneticists and other experts on CMT. Lunch is included in the registration fee for each conference. All speakers include time in their presentations for questions from the audience. Each conference begins with registration at 9:15am and concludes around 4:00pm. The all-inclusive cost for each conference is \$20.00.

Fill out the registration form below and return to the office of the CMTA by the date specified on the order form. Please mark carefully which conference you are registering for.



Houston or Akron CMT Conference Registration

Name(s) of attendees _____

Address _____

Telephone Number _____

Fee for one person \$ 20.00

(Fees include morning coffee, lunch, and all postage and mailings.)

We are attending the Akron conference

We are attending the Houston conference

Number of persons attending _____

Amount of money enclosed \$ _____

Return this form by September 24, 1993 for the Houston conference

October 22, 1993 for the Akron conference

To: Pat Dreibelbis
CMTA
601 Upland Ave.
Upland, PA 19015

Siblings... cont'd

Withholding information from siblings on the grounds that they are too young to understand only backfires, Perrin says, causing them to act out because they feel left out. Perrin is head of general pediatrics at Massachusetts General Hospital and chair of the American Academy of Pediatrics' Committee on Children with Disabilities. He has done extensive research on how families adapt to having a child with a chronic illness.

From the onset of an illness, children of all ages need concrete information at a level they can understand, according to DeMaso, including the real name of the disease.

To a three year old, you might say, "Your sister has juvenile rheumatoid arthritis. That means her fingers and her knees don't always work so well. They hurt sometimes. The medicines make them not hurt so much."

As a child gets older, and depending on his curiosity, more details can be shared, always at a level he can understand. To a 7-year-old: "Juvenile rheumatoid arthritis is a disease that causes joints not to work well. It's hard to predict when they will be painful, or why it happens, but all these medicines are helping her."

One of the most important things children under 11 need to know is that they did not cause their sister or brother to get sick. Blaming themselves is such a common thought among young children that pediatrician Betsy Busch says parents should assume it's going through a child's mind.

"They need to be specifically told, 'This is not your fault. You didn't make this happen,'" she says. And you need to say it repeatedly. Busch works with chronically ill children at the Floating Hospital at New England Medical Center.

Children as young as 2 and as old as 13 tend to equate love with the amount of time a parent spends with a child, according to pediatrician Ellen Perrin, an associate professor of pediatrics at UMass Medical Center in Worcester. Her private practice in Brookline specializes in children with chronic illnesses.

Thus, another typical thought might go like this: "You spend so much time with Amy, you must love her more." Perrin says this needs to be addressed head-on, too: "I spend a lot of time with Amy because her diabetes requires a lot of attention. That may make it seem like I

love her more, but I don't. I love you both the same."

It's not uncommon for school-age children to be angry at the sick sibling for consuming so much of a parent's time, and even to wish, as Todd Roy sometimes does, that she didn't exist. These thoughts can lead to feelings of guilt and jealousy, says Busch. Which is why parents need to convey over and over again that these emotions are OK to have and OK to talk about.

DeMaso suggests labeling and identifying feelings for young children. For instance, to a 3-year-old with a hospitalized sibling: "I know you must worry about your sister because she is sick, and you must be angry that Mama isn't home more often. It's okay to have that feeling."

In times of crisis, what can be difficult for siblings is not only the inaccessibility of a parent, which translates as a loss to a child, but also the disruption of regular routines.

"Give them the 'whys,'" says Busch. "They are more likely to feel helpful and proud to make a contribution if they understand why daily routines are being changed, why life has to revolve around the sib's therapy."

During a crisis, flexibility helps a family cope. While flexibility can mean some relaxing of limits for your healthy child-letting him stay up late so you can have some time together, for instance- it shouldn't mean excusing inappropriate behavior, says Busch.

For instance, if a sibling acts out by being mean or hostile, "You have to intervene," says DeMaso. "In our family we don't say mean things to each other," or "We don't shut the door on people, that's just rude behavior" and follow up with appropriate discipline. At a calmer time, he says, talk about the feelings behind the behavior: "I think you did that because you're upset with your brother. I wonder if you think he gets more attention than you do..."

In addition to meanness, there are two other troublesome behaviors that typically crop up among siblings:

- Being embarrassed by the sister or brother who is ill.

If this happens, DeMaso says, parents need to send a message that says, "This is our family, this is what we are." Do that, he adds, by encouraging a sibling to



bring friends home. If the sick child's behavior makes play difficult, he suggests structuring time when you can occupy your ill child. If there's equipment around the house, explain it matter-of-factly: "Tom's sister has trouble breathing. This machine helps her."

- Insisting that the sick child is faking it.

In this case, it's equally common for parents to come down on the sibling for thinking that. Instead, says DeMaso, examine the situation. "It may be true that she's faking," he says. "Then you need to say, 'I can see that you're right, but that doesn't give you the right to shut the door on her.' If it's not true, he says, it may be that the sibling needs more cognitive information. 'I know that it looks like her fingers should work. Some days they do, and some days they don't. We can't tell by looking which days they are hurting.'"

Todd Roy, who often complains his sister is faking it, says he knows, deep down inside, that she isn't. "I know she's really sick. I feel sorry for her. I really love her."

But, sometimes, he says, he just can't help his feelings.

Helping Kids with a Sibling's Illness

Always have contingency plans worked out and talked about in advance so that when a crisis pops up, siblings don't feel as though what happens to them is an afterthought.

Siblings tend to have a harder time when there is equipment in the home or when a professional (like a physical therapist) comes to the house regularly. Explain in simple language what the machines and people do.

Involving siblings in the care of their sister or brother can help them feel competent and valuable. Even a 3-year-old can tear tape or fetch items. But don't force involvement on them.

It's okay to cry in front of your children as long as you don't do it often - it can even be a model for them to handle their

(concluded on next page)

Siblings

feelings: "I feel so sad this is happening to your sister. Sometimes it makes me cry."

It's okay to express your frustration at not being able to spend more time with them. "I wish I could spend more time with you, but right now your sister is having an especially hard time. Once the new medicine (or surgery or therapy) starts to help her, I'll have more time for you. What can we plan to do together?"

Don't feel as though you always need answers for your children about their sibling. Just listening is important.

The child who cares for the sick sibling, who shows no jealousy or resentment and is always helpful, could be bottling up feelings. Give her openings to talk about them. §



Lungs - cont'd from p.12

phragm) motor latencies were abnormally prolonged, there was no correlation with the PFTs and airway pressure measurements.

Only 14% of 105 individuals had a history of frequent respiratory complications such as pneumonia or acute respiratory failure. Age, disease duration, spine deformity, pulmonary function, and phrenic nerve involvement did not significantly affect the incidence of respiratory dysfunction.

The relatively mild and essentially non-progressive impairment in pulmonary function and the low frequency of respiratory complications indicates that restrictive lung disease is usually not a major problem in HMSN (CMT). Since both the limb, back and expiratory musculature (as measured by MEP) are skeletal muscles, similar mild and slow progression of loss of strength would be expected in all three areas, but apparently with greater weakness in the latter. A reduced MEP is only rarely a cause of respiratory difficulty, since expiration is usually a passive action (unless complications arise from retained secretions secondary to an ineffective cough.) §

CMTA Membership/Order Form

Name: _____

Address: _____

Phone Number: _____

Tell us about yourself:

- CMT Patient
- Interested Supporter
- Medical Professional
- CMT Family Member

Enclosed is:

- \$25
- \$100
- \$50
- other

for my membership in the CMTA (newsletter included in membership)

- At this time I cannot contribute to the CMTA but would like to receive mailings.

Publications and Tapes available from the CMTA

(Check to order)

- VCR Tape - CMT Neurology (\$15)
- VCR Tape - Physical Therapy & Occupational Therapy (\$15)
- VCR Tape - CMT Genetics (\$15)
- VCR Tape - Orthopedic Surgery & CMT (\$15)
- Booklet (16 pp.) - CMT FACTS I (\$3)
- Booklet (24 pp.) - CMT FACTS II (\$5)
- Transcript - San Francisco CMT Conference (\$5)
- Letter - to Medical Professionals regarding the drug list (free to members with self addressed stamped business envelope)
- List - Physician Referrals (by state) (free to members with self addressed stamped business envelope) please list states: _____
- Medical Brochure - CMT (gray brochure) (free to members with self addressed stamped business envelope)

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

Total amount enclosed: _____

A copy of the official registration and financial information may be obtained from the Pennsylvania Department of State by calling, toll-free, within Pennsylvania, 1-800-732-0999. Registration does not imply endorsement.

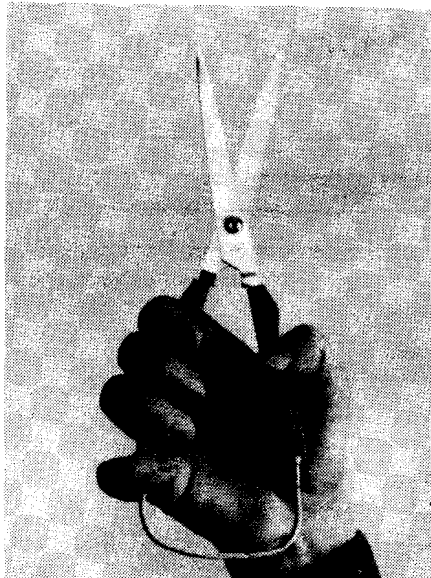
Call for Articles

The CMTA Report welcomes your ideas and article suggestions. For example, you may submit a human interest story telling of your experience of living with CMT. Also, medical professionals can forward articles of a clinical or medical nature that would be of general interest to our readership.

Do you know of anyone famous who is a CMT patient? Do you know anyone famous who would advocate for CMT?



The CMTA is looking for a well-known person to be a spokesperson for CMT. If you know of such a person, contact the CMTA; we will do the rest.



6. Loop Scissors: These are light-weight and spring loaded - you squeeze them to cut, the spring opens them when you open your hand. The nylon handles are easy to grasp. The blades are stainless steel.

Susan's Top Six - cont'd from p.7

ervation techniques. If you are followed by an MDA sponsored clinic for your CMT, your doctor can refer you to an occupational therapist as part of your clinic visit.

All of these items are readily available from medical suppliers. Listed here are some nation-wide medical suppliers who will be happy to send you their catalogs free of charge when you phone their toll free numbers. Difference in price among suppliers is usually very small, but postage is a consideration. Check the delivery charges and return policies prior to ordering.

Smith and Nephew Rolyan
1-800-558-8633

Fred Sammons, Inc.
1-800-323-5547

Cleo, Inc
1-800-321-0595
in Cleveland: 382-9700
in Ohio: 1-800-222-CLEO

Alimed
1-800-225-2610
1-617-329-2900

ABLEDATA
ask for the index of products available to assist in daily living
1-800-344-5405

Maneuverability
gadgets, fixtures, and appliances
1-800-522-1213

Bruce Medical Supply
1-800-225-8446

The author, Susan Salzberg, is employed at the Durham VA Medical Center in Durham, NC. The photographs were taken by the Durham VA Medical Media Department. The photographers were: Chris Baroody, Luke Thompson, and Ken Holt.) §

CMT...

- is the most common inherited neuropathy, affecting approximately 125,000 Americans.
- is also known as peroneal muscular atrophy and hereditary motor sensory neuropathy.
- is slowly progressive, causing deterioration of peripheral nerves which control sensory information and muscle function of the foot/lower leg and hand/forearm.
- causes degeneration of peroneal muscles (located on the front of the leg below the knee).
- causes foot-drop walking gait, foot bone abnormalities, high arches and hammer toes, problems with balance, problems with hand function, occasional lower leg and forearm muscle cramping, loss of some normal reflexes, occasional partial sight and/or hearing loss problems and scoliosis (curvature of the spine) is sometimes present.
- does not affect life expectancy.
- has no effective treatment, although physical therapy, occupational therapy and moderate physical activity are beneficial.
- is sometimes surgically treated.
- is usually inherited in an autosomal dominant pattern.
- may become worse if certain neurotoxic drugs are taken.
- can vary greatly in severity, even within the same family.
- can, in rare instances, cause severe disability
- is the focus of significant genetic research, bringing us closer to answering the CMT enigma.

THE CMTA REPORT

information on Charcot-Marie-Tooth disease from the

Charcot-Marie-Tooth Association

Crozer Mills Enterprise Center
601 Upland Avenue
Upland, PA 19015

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