JANUARY/FEBRUARY
2004

ISBN #1067-0181 ✲ Vol. 19, No. 1

THE CMTA Report

Information on Charcot-Marie-Tooth Disorders for patients, families, and the scientific community ✲ www.charcot-marie-tooth.org

Two New Grant Recipients Announced

The Charcot-Marie-Tooth Association has chosen to award $35,000 grants to two postdoctoral fellows beginning in January, 2004 and ending in December, 2004. Each works in the laboratory of a member of the CMTA's Medical Advisory Board.

Dr. Yan Huang is the recipient of the CMTA's Ann Lee Beyer Research Fellowship and will work with Dr. Steven Scherer of the University of Pennsylvania. Dr. Huang received her PhD from Fudan University in Shanghai and is extremely experienced in molecular biology. This project will capitalize on her molecular training but also extend those talents to the molecular pathogenesis of demyelination.

She will study the mutations in connexin32 that cause X-linked recessive CMTX1. This gene facilitates transport between the adjoining layers of the myelin sheath around axons and between the myelin sheath and the axon itself. Connexins form a large family and some are relevant to heart disease. Cx29 is also expressed in the nervous system, and, in particular, in Schwann cells. The function of Cx29 is unknown and no disease is associated with it.

The goal of Dr. Patino's study is to one day be able to treat human fetuses who have neurological disease.

Dr. Huang plans to characterize the function and properties of Cx29 by swapping domains between Cx29 and Cx32. This is a powerful technique used with a lot of success for many years. It will allow us to better understand the properties of Cx29 and Cx32 and likely identify other proteins with which Cx29 interacts. This study will lead to a better understanding of CMT in general.

The second recipient is Dr. Willmar Patino, working with Dr. Roger Lebo at the Children's Hospital Medical Center of Akron, Ohio. His fellowship is entitled, "Surgical Gene Therapy to Fetal Peripheral Nerves."

The goal of Dr. Patino's study is to one day be able to treat human fetuses who have neurological disease, obviously a dream for families and researchers alike. The challenge is to develop molecular techniques that will deliver the healthy copy of the disease gene into the particular cell type

(continued on page 3)

The CMTA Report Gets a New Look

Having just celebrated twenty years of service to the CMT community, the association thought that a new look for the newsletter would be in order. We hope you enjoy the new crisp, modern appearance of the report.

Many people have asked us to put the issue date on each page and we have responded to that request. Our goal is to make the newsletter easier to read and more polished as we head into our twenty-first year of operation. ✲

INSIDE:

CMTA
Charcot-Marie-Tooth Association

OUR MISSION:
To generate the resources to find a cure, to create awareness, and to improve the quality of life for those affected by Charcot-Marie-Tooth.

OUR VISION:
A world without CMT.

EMAIL CMTA AT:
CMTAssoc@aol.com

A REPORT FROM A 2003 GRANT RECIPIENT

Genotype-Phenotype Analysis of Mutations in the Human Neurofilament Light Gene Linked to Charcot-Marie-Tooth Neuropathy

RAUL PEREZ-OLLE, MD, PhD, Columbia University College of Physicians and Surgeons

During the year 2003, we made significant advances in our understanding of how mutations in the human neurofilament light (hNFL) gene cause Charcot-Marie-Tooth (CMT) disease. The results of our work, detailed below, have been submitted or are being prepared for publication.

One of the main aims of our studies has been trying to understand why patients with hNFL mutations have different clinical presentations. For this purpose we have investigated six new hNFL mutations, of which one appears to be a rare polymorphism not involved in the development of CMT. Analysis of the other five mutations has confirmed our observations with the first hNFL mutations described in CMT patients. All these hNFL mutant proteins cause alterations in the assembly of the intermediate filament (IF) networks, which are one of the main components of the "skeleton" of the nerve cells. We have discovered that some of the mutations have an increased tendency to form aggregates of mutant proteins. Overall, these results help explain why some CMT patients with hNFL mutations have almost normal nerve conduction velocities (NCVs) and are classified clinically as CMT-2E, while in other families patients have drastically reduced NCVs and are classified as CMT-1F.

The second main focus of our experiments has been determining in cultured neurons the effects of mutant hNFL proteins. We have observed that all hNFL mutant proteins are inefficiently transported and distributed into the axons, which are the very long processes of neurons that are responsible for transmission of the nerve impulses. Moreover, the mutant proteins also affect the distribution of a number of other proteins into the axons. Together, these results suggest that the hNFL mutations found in CMT patients cause generalized defects in the transport of neuronal proteins into the axons, analogous to traffic jams. We again observed that different mutant proteins resulted in slightly different effects, as previously described for the effects of hNFL mutations in the assembly of filaments.

The data we obtained in this fellowship year provide very interesting insights into the mechanisms by which CMT develops in patients with hNFL mutations. Hopefully, the knowledge gained will help explain how the disease develops in patients with mutations in other genes. The cell culture systems developed during the course of our studies should also be helpful in the investigation of potential therapies for CMT. ✲

The Non-Elderly Disabled

FROM THE NORD ON-LINE BULLETIN, JANUARY 2004

The Kaiser Family Foundation recently issued a study showing that non-elderly people who are receiving Social Security Disability Insurance (SSDI) are falling through the gaps in Medicare coverage. At least five million Americans are receiving SSDI benefits. After two years, they are eligible to receive Medicare.

However, the adult disabled population is generally very needy and has high health care needs. Because Medicare is designed primarily for elderly beneficiaries, it requires people to share some of the costs for medical products and services. Studies have shown that the disabled are twice as likely to live under the federal poverty level; they spend 40 to 50 percent

(continued on page 11)

NEW GRANTS

(Continued from page 1)

where it is needed and have it express there for the life of the individual. A common approach is to embed the gene in question into a virus because viruses have mechanisms that allow them to persist in a cell. These virus models are deprived of their disease-causing genes, but they still make proteins that our bodies recognize as foreign and that we fight with our immune systems. Therefore, engineered viruses that express healthy genes are often rejected after a couple of weeks by the body, and gene therapy fails.

Another challenge is to deliver the virus expressing the healthy gene into the correct cell type. The applicant wants to inject their constructs into the spinal fluid, where they would be taken up in dividing Schwann cells along nerve roots and hopefully these cells would express the normal gene from the virus and correct the abnormal phenotype.

Multiple different viral constructs will be tested and the virus will be injected in many different ways. The approach is obviously fascinating, but any human application is likely 5 to 10 years away. This approach would, predictably, be useful for CMT and many other conditions. ✲

Editor's note: We thank Dr. Florian Thomas, Chairman of the CMTA's MAB Grant Review Committee, for his explanations of the two grants.

CMTA MEMBERSHIP/ORDER FORM

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Phone Number: _____ Email: _____

Members who are current with their dues are considered "active." *If you are unsure as to whether you are current with your member dues, please call the office at 1-800-606-CMTA.*

	QTY	COST	TOTAL
Charcot-Marie-Tooth Disorders: A Handbook for Primary Care Physicians		*active members* $15 ***inactive members* $20**	
Membership Dues		$40	
CMT Facts I ☐ English ☐ Spanish		*active members* $3 ***inactive members* $5**	
CMT Facts II ☐ English ☐ Spanish		*active members* $5 ***inactive members* $7**	
CMT Facts III		*active members* $5 ***inactive members* $7**	
CMT Facts IV		*active members* $8 ***inactive members* $10**	
CMT Facts V		*active members* $12 ***inactive members* $15**	
A Guide About Genetics for the CMT Patient *No shipping and handling on this item only.*		*active members* $4 ***inactive members* $5**	
Golf Shirt *Size:* ☐ M ☐ L ☐ XL ☐ XXL		$15	
CMT Informational Brochure ☐ English ☐ Spanish		FREE	
Physician Referral List: States: _____ _____ _____		FREE	
Letter to Medical Professional with Drug List		FREE	
Contribution to CMT Research *10% will be applied to administrative expenses.*			
Shipping & Handling *Orders under $10 add $1.50, orders $10 and over add $4.50*			
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NIH Establishes Rare Diseases Clinical Research Network

To address the challenges inherent in diagnosing and treating rare diseases, the National Institutes of Health (NIH) announced the establishment of the Rare Diseases Clinical Research Network. With $51 million in grant funding over five years from several NIH components,* the network will consist of seven Rare Diseases Clinical Research Centers (RDCRCs), and a Data and Technology Coordinating Center (DTCC).

"Funding research on rare diseases is a vital aspect of the NIH mission," said NIH Director, Elias A. Zerhouni, MD. "By encouraging cooperative partnerships among the investigators at these centers, we hope to accelerate the development of diagnostics and treatments that will benefit these important patients."

The RDCRCs and the DTCC will be located at the following institutions:

- Baylor College of Medicine, Houston TX: Rare Disease Clinical Research Center for New Therapies and New Diagnostics (Dr. Arthur L. Beaudet).
- Boston University School of Medicine, Boston MA: Vasculitis Clinical Research Network (Dr. Peter A. Merkel).
- Children's Hospital Medical Center, Cincinnati OH: Rare Lung Diseases Clinical Research Network (Dr. Bruce C. Trapnell).
- Children's National Medical Center, Washington DC: Rare Diseases Clinical Research Center for Urea Cycle Disorders (Dr. Mark L. Batshaw).
- The Cleveland Clinic Foundation, Cleveland OH: Bone Marrow Failure Clinical Research Center (Dr. Jaroslaw P. Maciejewski).
- University of Rochester, Rochester NY: Nervous System Channelopathies Pathogenesis and Treatment (Dr. Robert C. Griggs).
- Weill Medical College of Cornell University, New York NY: The Natural History of Rare Genetic Steroid Disorders (Dr. Maria I. New).
- H. Lee Moffitt Cancer Center and Research Institute, University of South Florida, Tampa FL: The Data and Technology Coordinating Center (Dr. Jeffrey P. Krischer).

Approximately 25 million people in the United States are affected by an estimated 6,000 rare diseases or conditions. Diseases to be studies in the centers include: urea cycle disorders; Angelman's syndrome; Prader-Willi syndrome; Rett syndrome; periodic paralysis; non-dystrophic myotonic disorders; episodic ataxia; aplastic anemia; paroxysmal nocturnal hemoglobinuria; single lineage cytopenias, including granular lymphocyte leukemia, pure red cell aplasia, and myelodysplastic syndromes; vasculitis disorders; inborn defects in steroid hormone pathways; alpha-1 antitrypsin deficiency; lymphangioleiomyomatosis; pulmonary alveolar proteinosis; and hereditary idiopathic pulmonary fibrosis.

"Because of the small number of affected patients in any one location, rare disease research requires the collaboration of scientists from multiple disciplines and the capacity to share access to geographically distributed national research resources and patient populations," said Judith L. Vaitukaitis, MD, Director of the National Center for Research Resources (NCRR), one of the NIH funding components. "With a collaborative approach, the network will focus on identifying biomarkers for disease risk, disease severity and activity, and clinical outcome, while encouraging development of new approaches to the diagnosis, prevention, and treatment of rare diseases."

* The Rare Diseases Clinical Research Network is funded by the Office of Rare Diseases, National Center for Research Resources, National Institute of Child Health and Human Development, National Institute of Neurological Disorders and Stroke, National Institute of Arthritis and Musculoskeletal and Skin Diseases, and National Institute of Diabetes and Digestive and Kidney Diseases, all components of NIH, an agency of the Department of Health and Human Services.

OF INTEREST: Household Products Database

The National Institutes of Health (NIH) recently unveiled an online consumer's guide providing information on the potential health effects of more than 2,000 ingredients contained in more than 4,000 common household products. Known as the National Library of Medicine's (NLM) Household Products Database, it may be accessed at http://household-products.nlm.nih.gov.

"The network will facilitate increased collaboration and data sharing between investigators and patient support groups working to improve the lives of those affected by these diseases, and potentially prevent or eliminate these diseases in the future," said Stephen Groft, PharmD, Director of NIH's Office of Rare Diseases. "In addition, knowledge about rare diseases may offer leads for scientific advancement in other rare diseases and in more common diseases."

Each RDCRC will utilize the resources available at the General Clinical Research Centers—82 facilities distributed across the United States that provide clinical investigators with specialized research environments and specially trained research personnel. Supported by NCRR, the facilities include nursing staff, research subject advocates, and various core technologies including sophisticated laboratories, nutrition staff, and imaging facilities.

The creation of the network responds to the *Rare Disease Act of 2002*, which directed NIH to support "regional centers of excellence for clinical research into, training in, and demonstration of diagnostic, prevention, control, and treatment methods for rare diseases." The term "rare (or orphan) disease," as defined in the *Orphan Drug Act*, is a condition affecting fewer than 200,000 people in the United States, or a disease with a greater prevalence but for which no reasonable expectation exists that the costs of developing or distributing a treatment can be recovered from the sale of the drug in the United States. ✲

More Helpful Items

These items were found in Support Plus, a catalogue of medical helps. They can be purchased at 1-800-229-2910 or at www.supportplus.com

The Lori Shoe for Women
This is a versatile style that has a wider base outsole for added comfort and control. A hidden Velcro closure creates a wide opening and the Lori has extra depth and a removable insole that creates extra room for custom orthotics. Soft Cabretta leather, in navy, black, and wine. *$99.95.*

Easy Grips for your Door or Faucet
The deeply grooved rubber grips stretch over most doorknobs or faucets to allow easy turning. *$8.95 a pair.*

Yaktrax Walker
Yaktrax give you solid, predictable grip on packed snow and ice. They are made from injection molded plastic with steel wire coils that are hand wound in opposite directions to enhance stability. They are lightweight and easy to put on and take off. Available by shoe size. *$17.95 per pair.*

Good Grips Jar Opener
This unique jar opener works on any jar lid with only a twist of the wrist. Ideal for those with limited strength. *$10.95 (also available in kitchen stores and many department stores).*

DO YOU HAVE AN ITEM TO RECOMMEND?
Email CMTAssoc@aol.com

Understanding and Utilizing the Student IEP

A GUIDE FOR PARENTS NAVIGATING THE EDUCATION SYSTEM

The IDEA, the Individuals with Disabilities Education Act, is the federal law that governs all public school systems. The IDEA guarantees your child, from birth through age 21, a Free Appropriate Public Education (FAPE) in the Least Restrictive Environment (LRE). In other words, the school district legally must provide an educational program suited to your child's specific needs, in a setting as inclusive as possible.

The key tool for getting your child's FAPE is the IEP, an Individualized Education Program (or Plan). The IDEA mandates that each disabled child have an IEP and that the parents be involved in developing the plan.

By law, the district must call a meeting between you and school personnel to review your child's IEP at least once a year, but a parent, teacher, or administrator can call an IEP meeting at any time.

In some districts, the group that draws up the IEP is called the IEP team; other districts call it the Admission, Review and Dismissal (ARD) Committee. Generally it must include a school administrator, the child's special education teacher, the regular classroom teacher, personnel who've assessed your child's abilities, and one or both parents.

You may invite anyone else you wish to the IEP meeting; for instance, your child's physical therapist or doctor. By junior high, you'll also want your child to attend and have input. The law says children 16 and older must be invited.

Knowing your child's rights is crucial in formulating a specialized education program.

At the IEP meeting, the team members work out goals and objectives for the upcoming school year. They write an IEP that spells out exactly how those goals will be addressed. The plan should also include details about supports—whatever is necessary to enable the child to meet IEP goals and receive an appropriate education. This may include personal aides, a physical therapy program, adaptive computers, building modifications, note takers in class, and other needs. Failure to provide supports listed in the IEP violates the law.

An IEP is truly a group effort. The district, teachers, and family must agree on the final plan. Once it's put in writing and signed by you and the school, the district is legally obligated to provide whatever the IEP calls for—at no additional cost to you.

The IEP is a powerful document for students and parents. It means that you—as the person who knows your child's physical, intellectual, and emotional needs best—can insist that the school build his or her day around those needs. It means the school doesn't decide how to educate or accommodate the student; the school must work with you and be accountable to you.

The IEP process also allows you to take full advantage of

the school's expertise and resources. For the process to work at its best, all parties need to be both open to possibilities and realistic.

Before you go to an IEP meeting, check with your school or district's special education director or pupil service director for information. Most districts will give you a written set of guidelines and rules, including what to do if the committee can't reach consensus.

Be prepared to explain your child's physical condition; a written or oral report from the child's doctor can help. The IEP team may not be familiar with a neuromuscular disease, and this information can help clarify why modifications and assistance are needed.

You should also have a clear idea of what you want the IEP to accomplish.

Before the IEP meeting, examine the layout of your child's school, especially when he or she is going into a new school. See what changes are needed to provide accessibility to all areas.

If your child has a learning disability in addition to the neuromuscular disease, you'll need to prepare to focus on learning goals, as well as physical adaptations.

Think ahead of time about whether full inclusion is right for your child. Most of the time, inclusion has tremendous benefits for kids with physical disabilities. It raises the child's expectations, improves behavior and social skills, and makes the child feel part of the community.

But, in a few cases, a child with extreme fatigue, hearing or vision loss, or other additional problems may be too "medically fragile" to withstand a full school day. After consulting with your physician, you may decide that part-time or full-time home schooling would be the least restrictive environment for your child's education. Your team will draw up an IEP with academic expectations, tutoring requirements, and other details, all of which must be provided by the district.

The IEP should include a classroom modification page, listing such items as how the student will get to each class or activity. That may require the school to put in an elevator or provide an assistant.

> You should have a clear idea of what you want the IEP to accomplish.

Other modifications could be: a peer volunteer to take notes, extra time and an aide for test-taking, or modifications in the volume of work required. Your child may be given an instructional aide—a personal assistant to help in the restroom, push the wheelchair, and carry out physical therapy assignments.

For example, your IEP can specify having the child hooked up with a buddy during recess time, lunchtime, in the classroom; having a buddy help with their books and get them prepared for the beginning of class. For social and psychological reasons, classmates, not an adult aide, should assist the child with picking up items, handling books, and other simple classroom tasks.

The IEP can help older students learn self-advocacy and independence. It can require the student to explain his specific needs to teachers and ask for assistance. This is good practice in skills he'll need throughout life.

Parents are entitled to monitor how well the school district is carrying out the IEP.

If you're not satisfied that the school is complying with the IEP, keep records and document conversations about your concerns. If you can't work things out with the teacher, you can call an IEP meeting at any time to review progress.

All districts have a due process procedure to follow when members of an IEP team can't agree on program details. The steps generally first ask a district official to mediate. ✲

An excellent resource covering the IEP process in detail is You, Your Child, and "Special" Education: A Guide to Making the System Work *by Barbara Coyne Cutler (1993, Paul Brookes Publishing, (800) 638-3775).*

GIFTS WERE MADE

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CMTA REMEMBRANCES

Your gift to the CMTA can honor a living person or the memory of a friend or loved one. Acknowledgment cards will be mailed by the CMTA on your behalf. Donations are listed in the newsletter and are a wonderful way to keep someone's memory alive or to commemorate happy occasions like birthdays and anniversaries. They also make thoughtful thank you gifts. You can participate in the memorial and honorary gift program of the CMTA by completing the form below and faxing it with your credit card number and signature or mailing it with your check to: CMTA, 2700 Chestnut Parkway, Chester, PA 19013.

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In honor of (person you wish to honor)

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Send acknowledgment to:

Name: _____

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Occasion (if desired):

☐ Birthday ☐ Holiday ☐ Wedding
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Memorial Gift:
In memory of (name of deceased)

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Send acknowledgment to:

Name: _____

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Amount Enclosed: _____

☐ Check Enclosed ☐ VISA ☐ MasterCard

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Signature _____

Gift Given By:

Name: _____

Address: _____

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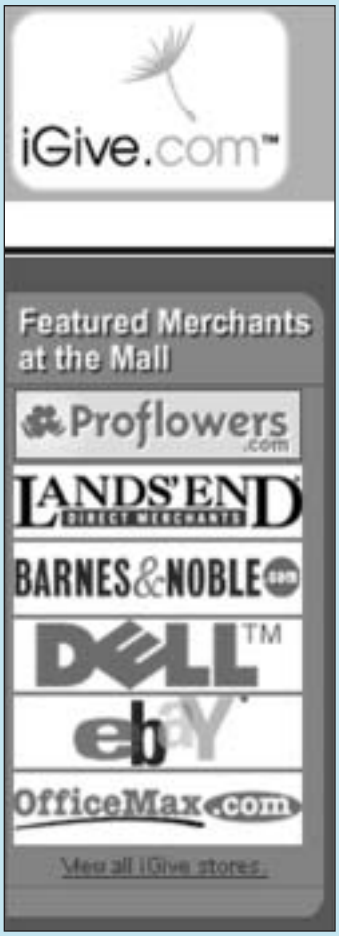
Other Ways to Give

In the past 12 months, the stock market has seen a positive northward movement. The Dow and S&P 500 are up approximately 20% and the NASDAQ is up approximately 50%. Many stocks are seeing their 12-month highs. Today, many investors have increasing value in their stock portfolios, mutual funds and 401ks. With CDs and money markets at historically low interest rates, the stock market has been a good alternative for many investors. However, unlike CDs and money markets, there are risks in stocks. There is, however, a risk-free investment that you can make. That investment is donating appreciated stock directly to the Charcot-Marie-Tooth Association. Investing in the CMTA is a powerful tool in supporting promising research and meaningful programs for CMT families.

Your investment in the CMTA, through appreciated stock, not only helps provide important programs, but it also allows you to receive significant tax advantages. Directly donating appreciated stock to the CMT enables you to make a tax deduction on the value of the stock donated and you will also escape liability for any capital gains tax. To take full advantage of the tax laws, you should contact your financial advisor or tax accountant before directly donating appreciated stock.

Another "painless" way to make a donation to the CMTA is by shopping on the Web. The two charitable shopping portals on our home page, www.GreaterGood.com and www.iGive.com, will take you to literally hundreds of stores. Each purchase you make at one of these stores will result in a percentage being given back to the CMTA. If you are looking for the latest fashions, books, office equipment, and more, you will find it on one of these shopping portals. So, if Santa forgot something you were hoping for, log on to the CMTA homepage and go shopping and know that some of what you spend finds its way back to the CMTA. ✶



When you shop at www.igive.com or www.GreaterGood.com, your purchases can support the CMTA.

IN HONOR OF (continued)

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William Shaw: Veteran, Research Hound, and Poet

Editor's note: I received a short letter from Hilda Studebaker suggesting that I run a piece on William Shaw because she and her brother knew him from their involvement with the clinic at Wayne State University, where Bill also has been treated. She suggested that he was an amazing man who continues, in spite of his CMT, to drag himself up on his tractor to mow his fields and who, as a side line, writes poems.

I wasn't surprised at all by her impression of Bill, since this office has known of Bill for years, and I'm happy to present him to our other readers.

William Shaw..."Cold Warrier" in more ways than one.

Bill Shaw was on active duty during the Vietnam war but escaped being sent there. He was a "cold warrior" and served as an AC&W (Aircraft Control and Warning), AKA radar operator, at a remote site on the US/Canadian border, where it was cold all the time. It was a small base, with the entire garrison, including support personnel, numbering around 140 airman, at most.

The men in operations watched the northern sky with our radar for the Russian Bear Bombers as they came up and over the Arctic Circle. The men tracked them as they flew down along the eastern seaboard past Boston, New York, Washington, and points south toward their ally, Cuba. The Cuban Missile Crisis happened on Bill's watch!

After his year of "remote," he spent two and a half years with the 26th Air Division in the Combat Operations Center at Hancock Field in upstate New York near Syracuse, where he did basically the same thing, air surveillance, weapons tech, and early warning operations, but never went to Vietnam.

The William Shaw that the CMTA office first knew was a man who knew more about the

TO "AMBULATION"

I would that my feet had the speed of my tongue,
I would still be quite active, lively and young.
But that's not the case for me here and now,
So my scooter I use, when I'm on the prowl.

Be it a mall in the suburbs, or at the State Fair,
My scooter works fine, I need not despair.
So quietly it glides through the stores in and out,
It's very dependable, really quite stout.

Red was my choice, for a real sporty color,
Its real nice to look at, and a very nice hauler.
I'd rather be walking, but that's just not the case,
But thanks to my scooter, I'm still in the race.

Will in KY

research being done on CMT than any lay person in the country. He assiduously pursued each and every laboratory that was doing any research even vaguely related to CMT and he wrote many letters encouraging CMT research even when the results were less than encouraging. When this office heard about some new and exciting research discovery, Bill was always, at least, one step ahead and had found out the same thing, by his own devices, before we had.

As for the poetry, Bill was characteristically shy about sharing it with our readers. He was finally convinced/coerced into agreeing that two poems wouldn't be such a huge step. The result is his "To Ambulation" and "A country place" which appear here in their entirety. ✲

A COUNTRY PLACE

Our place in the country, is quiet and serene,
Where the troubles of city life, out here are unseen.
Wildlife abounds all over this place,
And the scenes and the sounds, we wholeheartedly embrace.

Two deer just walked by, several minutes ago,
We see them quite often, from our windows, you know.
The variety of birds are too numerous to count,
We set out their feeders, for them to surmount.

The bees are all busy, making their honey,
The days seem pleasant, warm and quite sunny.
The color is green, the trees and the grass,
While butterflies abound, adding more class.

The ponds are teeming with fish; they swim and they float,
Where ducks and blue herons go fishing, because it's remote.
The occasional fox, whose den is concealed,
Runs and then plays, while he frolics in the field.

The landscaping is nice, with its shrubs and the flowers,
Made all the more pretty, with each passing shower.
And oh how the honeysuckle, with its fragrance so sweet,
Just grows everywhere, to give us a treat.

When late in July, the blackberries need picking,
We go out and get them, while our fingers we're licking.
All along the lane that leads to the pond,
Is filled with the berries, from there and beyond.

And then comes the autumn with colors so bright,
Followed by winter, with its blanket of white.
And with its rights of eminent domain,
Nature will renew itself, and start over again.

NON-ELDERLY DISABLED
(Continued from page 2)

more for prescription drugs; and they are twice as likely to spend more than 10 percent of their income on health care costs.

The Kaiser study found that almost half of the disabled population have an annual income under $12,000. About 60 percent reported postponing medical care due to costs, and 58 percent did not take their medicine as prescribed. Many have problems getting physician care, particularly because they can't find doctors who understand their disability. When they do find physicians, they tend to focus on the disabling condition and neglect normal health services. For example, fewer than half of disabled women had mammograms and only one third of men had prostate exams.

Advocates are calling on Congress to improve Medicare coverage for disabled people by eliminating the two-year waiting period before a disabled person qualifies for Medicare, helping people to get private insurance that could cover their deductibles and co-pays, and expanding the services that Medicare provides because:

- 46 percent of disability beneficiaries go without needed medical equipment or eyeglasses;
- 36 percent skip medication doses, split pills, or don't fill a prescription;
- 36 percent spend less on basic needs such as food or heat to pay for healthcare;
- 25 percent have trouble finding a doctor who understands their disability;
- 17 percent report a physician would not accept their insurance or Medicare;
- 15 percent have no regular doctor. ✲

SUPPORT GROUP NEWS

OF INTEREST: Drugs@FDA

The U.S. Food and Drug Administration (FDA) plans to roll out a new drug information Web site for consumers called Drugs@FDA. Simply enter Drugs@FDA in the search box on the FDA home page (www.fda.gov).

Consumers will be able to search by drug name and find out such things as whether a generic alternative of a product is available. FDA intends for the site to become a clearinghouse for drug-related regulatory histories, product labels, and manufacturing information.

Mississippi

The members of the support group were encouraged by the research that the CMTA is sponsoring for their disorder. They meet quarterly to discuss current research and how CMT is impacting their daily lives. They showed their support for the CMTA's efforts by donating $100 to the research fund. They also expressed their appreciation for publishing the findings so that they can better understand their illness and help other people in the future. This information was sent to the office by LaRue Wadford, Secretary of the group.

NW Ohio Support Group

The group met on Saturday, January 24, 2004 to hear group member, William Lloyd, speak on the topic, "Selecting Healthy Eating—Alternatively." William talked about maximizing the capacity that humans have to live very long lives. He quoted *The Guinness Book of Records* in saying that the oldest person lived to 122 in France and that the current oldest woman is in their state of Ohio, at age 114.

He stressed that aging comes from oxidation and stress and that the body replaces all its cells every seven years. He remarked that his body has changed, therefore, 11 times, even though it looks the same, only older. He closed his remarks with commentary on the advantages and disadvantages of white, as in white flour versus dark whole grains or the whites of eggs versus the yolks.

Pennsylvania, Johnstown

The Johnstown PA support group held its December meeting and Christmas Party at the Myron Williams Conference Room at Conemaugh Health System. The agenda included a presentation on bracing and orthotics by a representative from Svetz Orthotics & Prosthetics and Lori Murphy, PT, from Conemaugh. The members split into two groups and the presenters demonstrated a variety of AFOs, KFOs, and other braces. Judging from the animated discussions, this was an important subject. Much of the talk in our group concerned orthotic discomfort and finding shoes that will work with orthotics. Another subject of interest was insurance and MDA coverage for orthotics.

> Rosalie Grant has taken over leaadership of the State College group.

J.D. Griffith gave a brief PowerPoint presentation on recent research developments including Dr. Zarif Sahenk's success in treating CMT-1A patients at Ohio State with neurotrophin-3 and the work on progesterone in Europe.

Attendance was excellent and included Jan Goodard from Conemaugh and CMTA board member Pat Torchia.

We are excited about the Johnstown CMT Conference on May 26th at The University of Pittsburgh Living Learning Center hosted by The John P. Murtha Neuroscience and Pain Institute, an affiliate of Conemaugh Health System. The program will include a presentation by Dr. Michael Shy of Wayne State University.

We want to thank Conemaugh and the John P. Murtha Neuroscience and Pain Institute for their continued support.

Pennsylvania, State College

The State College CMT Support Group said goodbye to Katia Skovrinskie, who graduated from Penn State University this past winter. Although she does not have CMT, Katia adopted our cause as her own, not only starting the CMTA support group in State College, but organizing the meetings as well. The group would like to sincerely thank her for the great leadership and support.

Katia has chosen Rosalie Bryant, also a student at Penn State, to take over as support leader. The meetings are held monthly at the Centre Community Senior Center on Frasier Street in State College. Meetings usually fall on Thursday and last from 7:00pm until 9:00pm.

At the January meeting J. D. Griffith from the Johnstown group gave a PowerPoint presentation on recent CMT research developments. The February meeting will include a talk by a member of Conemaugh Health Systems.

The group is always looking for new members. If you would like more information, email Rosalie at rab296@psu.edu ✲

CMT Support Groups

Bob Budde, Support Group Liaison, 859-255-7471

Arkansas—Northwest Area
Place: Varies, Call for locations
Meeting: Quarterly. Meetings are not regularly scheduled so call ahead.
Contact: Libby Bond, 479-787-6115
Email: charnicoma57@yahoo.com

California—Berkeley Area
Place: Albany Library, Albany, CA
Meeting: Quarterly
Contact: Gail Whitehouse
Email: gwhite@earthlink.net

California—Northern Coast Counties (Marin, Mendocino, Solano, Sonoma)
Place: 300 Sovereign Lane, Santa Rosa
Meeting: Quarterly, Saturday, 1 PM
Contact: Freda Brown, 707-573-0181
Email: pcmobley@mac.com

Colorado—Denver Area
Place: Glory of God Lutheran Church Wheat Ridge
Meeting: Quarterly
Contact: Marilyn Munn Strand, 303-403-8318
Email: mmstrand@aol.com

Kentucky/Southern Indiana/ Southern Ohio
Place: Lexington Public Library, Northside Branch
Meeting: Quarterly
Contact: Martha Hall, 502-695-3338
Email: marteye@mis.net

Massachusetts—Boston Area
Place: Lahey-Hitchcock Clinic, Burlington, MA
Meeting: Call for schedule
Contact: David Prince, 978-667-9008
Email: baseball@ma.ultranet.com

Minnesota—Benson
Place: St. Mark's Lutheran Church
Meeting: Quarterly
Contact: Rosemary Mills, 320-567-2156

Minnesota—Twin Cities
Place: Call for location
Meeting: Quarterly
Contact: Maureen Horton, 651-690-2709
Bill Miller, 763-560-6654
Email: mphorton@qwest.net, wmiller7@msn.com

Mississippi/Louisiana
Place: Baptist Healthplex, 102 Clinton Parkway, Clinton, MS
Meeting: Quarterly
Contact: Flora Jones, 601-825-2258
Email: flojo4@aol.com

Missouri/Eastern Kansas
Place: Mid-America Rehab Hospital, Overland Park, KS
Meeting: First Saturday bimonthly
Contact: Lee Ann Borberg, 816-229-2614
Email: ardi5@aol.com

Missouri—St. Louis Area
Place: Saint Louis University Hospital
Meeting: Quarterly
Contact: Carole Haislip, 314-644-1664
Email: c.haislip@att.net

New York—Greater New York
Place: NYU Medical Center/ Rusk Institute, 400 E. 34th St.
Meeting: 2nd Thursday of each month
Contact: Dr. David Younger, 212-535-4314, Fax 212-535-6392
Website: www.cmtnyc.org

New York—Horseheads
Place: Horseheads Free Library on Main Street, Horseheads, NY
Meeting: Quarterly
Contact: Angela Piersimoni, 607-562-8823

New York (Westchester County)/ Connecticut (Fairfield)
Place: Blythedale Hospital
Meeting: 3rd Saturday of each month, excluding July & August
Contacts: Beverly Wurzel, 845-783-2815
Eileen Spell, 201-447-2183
Email: cranomat@frontiernet.net espell@optonline.net

North Carolina—Archdale/Triad
Place: Archdale Public Library
Meeting: Quarterly
Contact: Ellen (Nora) Burrow, 336-434-2383

North Carolina—Triangle Area (Raleigh, Durham, Chapel Hill)
Place: Church of the Reconciliation, Chapel Hill
Meeting: Quarterly
Contact: Susan Salzberg, 919-967-3118 (evenings)

Ohio—Greenville
Place: Church of the Brethren
Meeting: Fourth Thursday, April–October
Contact: Dot Cain, 937-548-3963
Email: Greenville-Ohio-CMT@woh.rr.com

Ohio—NW Ohio
Place: Medical College of Ohio
Meeting: Quarterly
Contact: Jay Budde, 419-445-2123 (evenings)
Email: jbudde@fm-bank.com

Oregon/Pacific NW
Place: Portland, Legacy Good Sam Hospital, odd months Brooks, Assembly of God Church, even months
Meeting: 3rd Saturday of the month (except June and Dec.)
Contact: Jeanie Porter, 503-591-9412 Darlene Weston, 503-245-8444
Email: jeanie4211@hotmail.com or blzerbabe@aol.com

Pennsylvania—Johnstown Area
Place: Crichton Center for Advanced Rehabilitation
Meeting: Bimonthly
Contact: J. D. Griffith, 814-539-2341
Email: jdgriffith@mail.charter.net

Pennsylvania—Northwestern Area
Place: Blasco Memorial Library
Meeting: Call for information
Contact: Joyce Steinkamp, 814-833-8495
Email: joyceanns@adelphia.net

Pennsylvania—Philadelphia Area
Place: Penn Towers Hotel Conference Room
Meeting: Bimonthly
Contact: Amanda Young, 215-222-6513
Email: stary1@bellatlantic.net

Pennsylvania—State College
Place: Centre County Senior Center
Meeting: Monthly
Contact: Rosalie Bryant
Email: rab296@psu.edu

On Stubbornness, Disability, and Making Do

PAT DREIBELBIS

I don't often write anything personal. As an editor, one is expected to stay somewhat invisible and removed from the articles that are published. On this occasion, however, my personal experiences are related to both my employment at the CMTA and my ability to relate to people who have CMT.

On November 16th, two days after my birthday, I was watching the Eagles on television and wrapping Christmas presents (yes, I'm annoyingly one of those people who get things done ahead of time and never, never wait until the last minute). At halftime, I decided to go outside and clean up some branches that had fallen during one of our horrible winter storms. I was doing just that when I found a branch that was too long to fit in my trash container. Knowing how persnickety the trash collectors can be, I decided to break the branch in half by standing on it and pulling the end of it up with my hands. Oblivious to the likely dangers in such a move, I stepped up on the "round" branch and pulled. My feet rolled with the branch and I was thrown backwards into the street, landing on my left elbow.

I knew from the moment of impact that something terrible had happened. The immediate problem was that I was lying on my back in the street and no one else was outside to see my plight. Fortunately, a few minutes later, my neighbor came out and responded to my feeble calling of his name. With some considerable effort, he helped me up and I went into my house, thinking and hoping that nothing was "really" wrong. Of course, it took only a moment to realize that something was terribly wrong because the pain was excruciating and movement of my left arm was NOT an option. I was taken, immediately, to the emergency room of the nearest hospital.

Ian Britton, courtesy of FreeFoto.com.

Trying to type with one hand was one of the challenges faced by CMTA editor Pat Dreibelbis after her accident.

I'll skip the details, painful and gory as they were, but I was x-rayed, found to have broken three bones in my elbow and told that surgery was likely my only option. I had a shot in my "haunch," and for a period of time felt no pain while they put a temporary cast on my arm and made a surgeon's appointment the following day. Three days later, I was in and out of surgery in no time and sent home with a massive cast on my left arm which covered all but the tips of my fingers. I had titanium bars and wires holding my arm together, and I will have them for the rest of my life.

The number of things that require two hands to do is probably well known by all of you readers. I was amazed at how cutting my food was impossible, how fastening a bra was absolutely out of the question and how tying shoelaces was a two-handed task. I learned humility when I had to journey from house to house in my neighborhood looking for someone to tie my shoes so I could safely walk around. (Since I'm diabetic, walking without shoes isn't really an option for me.) I couldn't curl my hair, let alone take a decent shower, so vanity was a trait I couldn't enjoy for weeks.

Within four days of surgery I was back at work at the CMTA but found that typing with only one hand was so slow as to be useless. We devised a plan where I printed out email questions and then dictated the answers to one of the other women in the office. I could still answer the phone and I did that. The December issue of the newsletter was just about finished and, again, I was able to get that done by dictating articles to others. Eventually, the first cast was replaced with a second one, which left my fingers out. Then, I returned to my own typing, even though it was still a slow and painful process.

The CMTA Report is published by the Charcot-Marie-Tooth Association, a registered non-profit 501(C)(3) health organization. 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.

More importantly to me, I could now, with some difficulty, go back to wrapping Christmas presents and making Christmas cookies (even though the surgeon discouraged these activities). To describe the manner in which these tasks were accomplished would be to somehow paint a picture of how someone with a very heavy cast on one arm can unroll paper, cut (really difficult with only one arm that works correctly), and then tape together the ragged edges into something resembling a package. The act of rolling out dough and cutting cookies was even more pathetic, but I got them done and my children were smart enough to say that they looked "pretty good, all things considered." I would suffer for hours after these activities, but I was too stubborn to let go of our Christmas traditions.

My cast was off by Christmas Day, but my left arm, even now, is still very weak and not capable of lifting and carrying anything of any weight. Consider then, the challenge of stuffing a 16 1/2 pound turkey and getting it and my mother's old enameled turkey cooker into the oven by myself. I'm not sure to this day exactly how I did it, but I know it involved embracing the turkey in something like a wrestling move and holding it up by one leg while I pushed as much stuffing as possible in its cavity.

Happily, my 6'4", 240 lb. son was home by the time it needed to come out and he could lift and carry the bird and its roasting pan without much effort. The meal was a success in spite of the difficulties involved and I have learned from all this that it doesn't hurt to be stubborn and to find alternative ways to do things when you are disabled in some way. I think it was a lesson I was meant to learn so that I can relate even more appropriately to the stories I hear of CMT patients who do amazing things in spite of their disabilities. ✲

LETTERS

To Whom it May Concern,
Enclosed please find a money order as a donation to your Association.

I have been a member of your association for many years. I have a daughter and a grandson with Charcot-Marie-Tooth and it is very difficult sometimes to see the trials they have to go through day after day. Your newsletter has supplied information and encouragement for us. I must say that your association has made great strides in research and hopefully will continue to make even further strides in the years to come to help people with this disease.

I sincerely thank everyone who is working within your association for their continued efforts to find a cure for this debilitating disease.

—S.A. Gardner, MA

Dear CMTA,
At the end of the day, my feet hurt and I can't wait to get my shoes off. But slippers are too soft and don't give me enough support. The solution that works for me is State Street Shoes #2828—which I found at Payless Shoe Source for $19.99. These slip-ons fit my very narrow feet fine. They offer good support, are comfortable and warm, and non-skid.

—From an email

Dear CMTA,
Thank you for supporting Dr. Willmar Patino as a CMTA Research Fellow to test the feasibility of delivering therapeutic genes to growing fetal nerves. This will help us determine whether thsi could be an effective approach to cure CMT.

Now that we and other scientists have found most of the CMT genes and developed diagnostic tests to categorize our patient's abnormalities unambiguously, the next step is to search for a cure. Gene therapy modulates unique gene expression and aims to correct different mutations specifically. We thank you for providing us with the resources not only to complete the next step in the process but to help position us for greater contributions.

We shall be proud to acknowledge the CMTA in any report that we submit for publication. We published a chapter on human chromosome centromeres that acknowledges the CMTA's previous support. Centromeres may prove to be an effective gene delivery vector in our prenatal studies.

—Roger V. Lebo, PhD, FACMG

WRITE TO US!

Pat Dreibelbis, Editor
The CMTA Report
CMTA
2700 Chestnut Pkwy.
Chester, PA 19013
or
CMTAssoc@aol.com

The CMTA reserves the right to edit letters for space.

MEDICAL ALERT:
These drugs are toxic to the peripheral nervous system and can be harmful to the CMT patient.

Adriamycin
Alcohol
Amiodarone
Chloramphenicol
Cisplatin
Dapsone
Diphenylhydantoin (Dilantin)
Disulfiram (Antabuse)
Glutethimide (Doriden)
Gold
Hydralazine (Apresoline)
Isoniazid (INH)
Megadose of vitamin A*
Megadose of vitamin D*
Megadose of vitamin B6* (Pyridoxine)
Metronidazole (Flagyl)
Nitrofurantoin (Furadantin, Macrodantin)
Nitrous oxide (chronic repeated inhalation)
Penicillin (large IV doses only)
Perhexiline (Pexid)
Taxol
Vincristine
Lithium, Misomidazole, and Zoloft can be used *with caution.*

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

*A megadose is defined as ten or more times the recommended daily allowance.

What is CMT?

- is the most common inherited neuropathy, affecting approximately 150,000 Americans.
- 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 also known as peroneal muscular atrophy and hereditary motor sensory neuropathy.
- is slowly progressive, causing deterioration of peripheral nerves that 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).
- does not affect life expectancy.
- 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, and scoliosis (curvature of the spine).
- 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, which means if one parent has CMT, there is a 50% chance of passing it on to each child.
- Types 1A, 1B, 1D (EGR2), 1X, HNPP, 2E, 4E, and 4F can now be diagnosed by a blood test.
- is the focus of significant genetic research, bringing us closer to answering the CMT enigma.

THE CMTA *Report*

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