FootSteps

Fall 2007
Vol. 8, No. 3

The newsletter for the members of The Erythromelalgia Association
***FootSteps* online: www.erythromelalgia.org**

Fundraising Is Challenging for TEA

Do you have a favorite—or not so favorite—charity that sends you one request for donations after another, and another, and another?

And at the end of the year, non-profit organizations send their biggest and best "annual appeals."

Except TEA.

But this fall, TEA hopes to send out an appeal for money to fund programs to help people with EM.

In addition to the Research Fund, TEA's board of directors last year started a General Fund.

The board set goals for TEA to fund more programs to help people with EM ***now*** using General Fund money.

These include educational programs like the 2006 teleconference with Dr. Cohen, for example.

And this year the board plans to do an update of the 2003 survey of members.

This 2003 survey provided facts about treatments that numerous people with EM have shared with their doctors during the past four years.

It's an example of a project that took hundreds of one TEA volunteer's hours to make happen.

This time TEA needs to hire a professional survey firm to do the work. And that costs money.

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Also In This Issue

Dr. Costa Enjoys Dobbs Interview

Editor's Note: "The Pain Gate" now is on TEA's Web site: erythromelalgia.org. Click on "In the News."

It was not just another media interview.

TEA member Pam Costa, Ph.D., was more than happy to talk to freelance writer David Dobbs because she knows Dobbs' work.

She requires her psychology students at Tacoma Community College, Tacoma, WA, U.S., to read Dobbs' articles in *Scientific American Mind.*

Dobbs is a frequent contributor to the magazine, which investigates and analyzes new thinking in psychology, neuroscience and related fields.

"We talked for two hours," Dr. Costa reports.

The resulting article tells Dr. Costa's story of living from infancy with the often excruciating pain of inherited EM. She's now 42.

"The Pain Gate," in the April-May 2007 issue of *Mind*, also tells the story of Stephen Waxman, M.D., Ph.D., chair of neurology at Yale University, and his neuroscience laboratory of researchers.

Dr. Costa belongs to a large family with many members affected by EM. They were studied by the Mayo Clinic in 1966 and University of Alabama in the 1991.

But it was not until many years later, Dobbs' article relates, that the rare, sometimes genetic disease EM came to the attention of pain researchers around the world and especially those at Yale.

The scientists at Yale's West Haven Veterans Administration Rehabilitation Re-

(Continued on page 6)

Q and A by Gayla Kanaster

Send answers and new questions to Gayla, 2532 N. Fremont St., Tacoma, WA, USA 98406, or GaylaKanaster@aol.com

NEW ANSWERS, OLD QUESTIONS

Q "What food and drinks, including caffeine, have an effect on EM?" (Nicki Greer)

A Stephanie Spaide, Boston, MA, U.S.: "I find that alcohol, primarily wine, causes immediate EM symptoms while hard liquor does not seem to have such a dramatic effect. As for food, I have recently begun to think that carbohydrates (pastas, breads, crackers, starchy vegetables) worsen my symptoms, while a lower carbohydrate, high protein diet does not cause as much swelling and burning in my hands and feet."

A Philip Singleton, Cornwall, UK: "Others have commented red wine and beer cause them to flare, but I drink one glass of red wine a day, as advised by my urologist."

A Pam Costa, University Place, WA, U.S.: "I cannot tolerate any kind of alcohol, even wine cooked in food. When I eat out, I've learned to ask if certain marinades or recipes have alcohol in them. Many times I'm told, 'yes—but the alcohol cooks out.' It still makes me burn. This is probably because alcohol is a vasodilator.

Q "Does anyone else wake up with pain and redness in their feet and legs after sleeping a short time?" (Pam Costa)

A Philip Singleton: "When standing first thing in the morning, I experience a large increase in redness, soreness, burning pain. I am not able to take beta blockers because of other medical problems. My G.P. also will not recommend amitriptyline. I have burning pain which never ceases and often wakes me up during the night, some nights worse than others. I take one-half Nitropam tablet when desperate to get sleep. My feet, legs, left hand and lumbar region of my back are affected."

Singleton adds that he "has been a TEA member for several years and turned 87 in August. The only local help/advice/information, etc., is from Jean Jeffery. We have frequent talks on the phone. She ... puts in so much work and effort on behalf of people with EM. (Thanks so much, Jean.) Also, thanks all at TEA for your time and dedication to help those of us with EM. "

NEW QUESTIONS

Q "Does anyone have any recommendations for those with EM and Raynaud's? I have neuropathy and some days it is so hard." (Submitted by **Tina Powell**)

Q "Has anyone tried Solaraze gel on the feet and legs? It's used for pre-cancers (actinic keratoses), and rheumatoid arthritis, sometimes with great results." (Submitted by **Allison Bachman,** Casselberry, FL, U.S.)

Q "Besides having EM in my feet and legs, I believe it has caused swelling in my face, tongue and lips, to the point that it causes me to lisp. Has anyone experienced that?" (Submitted by **Mary Moulton**, Sandy Spring, MD, U.S.)

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Families with EM Mutations Investigated

By Jean Jeffery

This report describes two papers [1,2] by Dr Joost Drenth about the EM gene in several families with inherited EM. The first paper can be found in TEA Articles (Research).
The faulty gene lies on chromosome two and makes the protein for the sodium 1.7 channel (Nav1.7). Sodium 1.7 channels are tiny openings along the surfaces of nerve fibers. Each channel is a complex structure containing four sets of "batteries" that control the flow of sodium ions into the nerves to send electrical signals. These messages are relayed from the pain-sensing nerves in the skin to the spinal cord and brain.

Five families studied

The five families with EM lived in France, The Netherlands, Belgium, and Canada. The burning pain of EM affecting their feet, or feet and legs, was dominantly inherited. This means that when one parent has the faulty gene, the risk of passing EM to each child is 50 % for each pregnancy. Of the two French families, one had five members from three generations affected, including two young boys with very severe EM. Painful attacks during the day and night were relieved by keeping their limbs in cold water. The second family had one parent and two young daughters with EM. A father and daughter in the Dutch family were severely afflicted with constant burning pain. Sadly, one member from this and the second French family died from septic ulcers caused by excessive cooling of their limbs in cold water. The Belgian family had ten individuals with EM from four generations. The most severely affected were two teenage boys who needed methadone and constant air-cooling to relieve their EM. In the Canadian family one mother and her three children had EM. The mother and son had mild symptoms whereas her two daughters were severely affected.

Mutations cause pain

Blood samples were taken from members of each family for DNA analysis. A mutation in the sodium 1.7 channel was found in all individuals with EM, but was absent in all individuals without EM. First symptoms of EM always began in childhood, between one and 10 years of age, and persisted through life. Each family had a different mutation within the sodium channel: the French families had the mutations I848T and F216S, the Dutch N395K, the Belgian S241T, and the Canadian family L858F.

The sodium channel protein is composed of precise sequences of amino-acids. A mutation changes the amino-acid sequence: thus in the French mutation I848T the amino-acid T: threonine replaces the amino-acid I: isoleucine; in the Belgian mutation S241T threonine replaces S: serine. These alterations in the protein disrupt the normal behaviour of the sodium channels to cause the painful symptoms of EM. When the mutant channels open, the sodium ions rush in and send high numbers of abnormal pain signals from the skin to the brain. Each mutation behaves in a slightly different way.

Treatment for one of the five families was partially successful with a single infusion of lidocaine followed by mexiletine. Four members in the French family with the I848T mutation experienced improvement in their symptoms for 1-2 years. Lidocaine and mexiletine are sodium channel -blocking drugs. It is hoped that a new drug will be found that specifically targets the 1.7 channel to alleviate EM.

Clarification: The June 2007 issue carried an article that summarizes two papers about treatment of EM patients with lidocaine skin patches. The description of the patch and its use should contain this warning about the potential misuse of these patches: ***A maximum of*** three patches *must* be worn ***only*** for up to 12 hours within any 24-hour period. (Follow your doctor's prescription.) If directions are followed, the lidocaine will penetrate the skin to reach damaged nerves, but not enter the bloodstream to cause any adverse effects.

[1] SCN9A mutations define primary erythermalgia as a neuropathic disorder of voltage-gated sodium channels. Drenth JP, Te Morsche RH, Guillet G, Taieb A, Kirby RL, Jansen JB. Journal of Investigative Dermatology 2005, 124:1333-1338.

[2] Autosomal dominant erythermalgia associated with a novel mutation in the voltage-gated sodium channel alpha subunit Nav 1.7. Michiels JJ, Te Morsche RH, Jansen JB, Drenth JP. Archives of Neurology 2005, 62:1487-1590.

Your Stories—everyone has one!

We can all empathize with fellow members who face the daily challenges of living with EM. Because EM is so rare, most of us have tales of the often long and difficult diagnosis process and the ways we've found to cope.

Sidsel Horgen Wert writes: I received my first *Footsteps* and would like to share my mother's story. Her name is Inger Horgen, she's 84, and lives in Tidaholm, Sweden.

Two years ago she began having a burning sensation on the bottom of her feet at night. They became red/blue and swollen, and she had to get up and cool her feet in cold water. It lasted a couple of hours every night and she had no idea what caused it.

I checked the Internet and found an article by Dr. Cato Mork, who is doing EM research in Norway. Her symptoms fit his description of EM. Her personal doctor had never heard of this condition, but put her on the medication Misoprostol (also called Cytotec in the U.S.) to dilate the blood vessels. Dr. Mork had tested it on some EM patients, but there was no improvement on my mother's part.

Last year she tried taking a protein drink, which contains vitamins, minerals and amino acids. After three weeks she had no problem with her feet and this continued for about five months. But the EM slowly returned.

In January she started getting acupuncture. She is pain free in periods and when the pain returns it is tolerable and she does not have to get up at night. I believe she will have to have acupuncture for as long as she lives. An important aspect about acupuncture, in my opinion, is to get treatment from a person with a high level of education. (In Scandinavia it is very popular to take short-term courses in acupuncture.)

John Ravetti writes: My daughter Danielle, who had been a Regional Roller Skating Champion and an avid basketball player, started experiencing pain so bad when she was 13 that she started walking on the outside of her feet. This led to a horrible gait problem.

Her doctor and podiatrist could not figure out what was wrong. A highly respected orthopedic surgeon said he could correct this gait problem by cutting the back of her heel. Another surgeon said that her flat feet were the contributor.

A physiatrist recommended intensive inpatient care for three to five weeks. After two days in the hospital she was diagnosed with Conversion Disorder and Complex Regional Pain Syndrome (CD/CRPS). We were told that some outside stressor caused this CD diagnosis.

As parents we were under an excruciating amount of stress. What outside source was causing my daughter this pain and abnormal gait? The worst pain a parent can experience is not knowing how to help your child. Many times I would wake up in the middle of the night because of my daughter's crying.

We were struggling for answers. We were referred to Shriner's Hospital, in Sacramento, where she was diagnosed with Reflex Sympathetic Dystrophy (RSD). I looked into the various symptoms of RSD and noticed that a couple did not match Danielle's.

Through the help of my aunt, we were fortunate to be referred to the Pain Management Team of Lucille Packard Children's Hospital at Stanford University in Palo Alto, CA. She was immediately diagnosed with EM. Danielle (and her parents) were relieved to finally have a diagnosis. It wasn't in her head.

The Pain Management Team closely evaluated her—all had a deep concern for her well-being. In her four-week stay she accomplished her goal of walking with crutches for

Your Stories—everyone has one!

TEA encourages you to share your experience by writing your story. We can help you write and edit your story. We ask only that you limit it to about 400 words. Please send it to Gayla Kanaster, gaylakanaster@aol.com or 2532 N. Fremont Street, Tacoma, WA, USA 98406

her eighth grade graduation. Several weeks later, the head doctor at Stanford met one of the physician pioneers of a pain treatment using capsaicin cream. This doctor had successfully performed the procedure for adults.

Three weeks later, Danielle was back in the children's hospital at Stanford to receive the treatment with 10 percent capsaicin cream. She had an epidural anesthetic during the procedure. The second day she complained of the fire on both the top and bottom of her feet.

On the third day she walked out with zero pain. It worked! She's been pain free for the last four weeks and has started high school. It had been more than 18 months since she had walked pain free with a normal gait.

In summary, I would like to tell all who are reading this to never give up. Never give up researching. You don't have to accept your doc-

Stephanie Spaide writes: I am 41 years old but my struggles with EM and other rare disorders began in my 30s. I have been diagnosed with Ehlers-Danlos syndrome, Raynaud's syndrome, peripheral neuropathy, and EM. It has been a long, sometimes exhausting and painful journey. The absence of symptoms (burning hot, red, itchy feet/hands) or often the presence of the opposite symptoms like ice cold, mottled hands and feet during doctor's appointments delayed my EM diagnosis. Photographs of flare-ups brought to appointments ultimately provided MDs with "evidence" to make the EM diagnosis.

Having EM and Raynaud's makes managing symptoms difficult. It is a delicate balance; what helps to alleviate one, aggravates the other. During Christmas, I stood on the cold sidewalk in my bare feet to alleviate the burning hot pain and swelling after a day of shopping. Later, I used a heating pad for the searing pain of Raynaud's. I have had Raynaud's symptoms in my left hand while simultaneously having EM symptoms in my right hand.

If given a choice, I would take the Raynaud's over the EM. Raynaud's leaves my hands/feet so cold they lose feeling while EM flare-ups cause my hands and feet to throb with burning heat and swelling. During an EM flare-up, I would do anything to make it hurt less, including icing. The dangers of ice became obvious when I got frostbite after icing a broken ankle!

Although initially devastating, a career change due to EM has proven beneficial to my health and happiness. I left medical sales to return to the field of nutrition. The more sedentary job has reduced my pain significantly, and I feel my health problems have made me a better clinician with tremendous empathy for my patients' struggles.

I recently began using the amitriptyline and Ketamine gel trialed at the Mayo Clinic. So far, it's the first treatment to provide me with relief!

There is no simple solution to my medical problems. My healthcare providers along with TEA provide me with the support, encouragement and hope I need to persevere through the tough times. It makes all the difference.

***Scientific American Mind* Article** *(Continued from page 1)*

search Center, under Dr. Waxman's direction, have been on a decades-long journey of their own—trying to understand the fundamental mechanisms of neuropathic pain with the aim of finding a cure.

An estimated 50 million people in the U.S. alone have some form of neuropathy or nerve pain, according to Dobbs' article.

These include diabetics, wounded soldiers, shingles patients, to name just a few. And people with EM.

When Dobbs learned about Yale's work with EM, he was "immediately interested." As were the editors of *Mind*.

TEA led him to Dr. Costa, who already had been interviewed by a *Tacoma News Tribune* reporter and the Discovery Health Channel.

And TEA had led Yale to Dr. Costa and the rest of her family, most of whom live in Alabama. They willingly provided blood samples in 2004.

The article explains what the researchers discovered using DNA from these samples.

Pam Costa, Ph.D.

And how they used 21st century methods of gene manipulation and observation to isolate mutations to an already suspect gene. Mutations they then linked to the pain of EM.

And how the scientists are still in the laboratory using EM to carefully learn the fundamental biology of neuropathic pain.

Dobbs was interested because EM is so rare "and Waxman's discovery clearly carried such great potential to eventually lead to relief not just for those with EM but for other sufferers of chronic pain."

Scientific American Mind is a spinoff of the venerable in-depth science news magazine *Scientific American*.

Dobbs also writes for the *New York Times Magazine*, among others. Read his work at http://daviddobbs.net.

Nicki Greer

New Board Member

Joining the TEA Board of Directors this summer was Nicki Greer of Aurora, Ohio, U.S.

A TEA member for just two years, Greer already volunteers her time assembling and sending packets of information to new members.

Greer has two grown children and two grandchildren. "My first priority has always been my family." she says. "My EM seems to be controlled right now, and for that I am grateful." Since her diagnosis, she has "made it her passion" to find out all she can about EM so she can "deal with the reality."

Mom Raises Money for Yale Research

An appeal e-mailed last March by TEA member Sharon Hoff, Briarcliff, N.Y., U.S., raised more than $70,000 for EM research.

Hoff wrote the e-mail after her then 11-year-old daughter Sydney was diagnosed with primary EM. Sydney now is one of the children being studied by the research team at Yale University's West Haven Veterans Administration Rehabilitation Research Center.

Hoff collected the donations herself (instead of sending them to TEA's Research Fund). She so far has personally given checks totaling $50,000 to Stephen Waxman, M.D., Ph.D., director of the center at Yale.

Sent to everyone in Sydney's school district, Hoff's e-mail described how her daughter suffered from the burning pain and how dramatically her life had changed since contracting EM.

She described the research being done at the Yale

Wedding Gift Registry Nets More Than $2,700 for TEA

When Todd and Karen Kanaster planned their August 25, 2007, wedding in Denver, CO, U.S., they decided to ask for gifts to TEA instead of presents.

They set up a wedding Web site with a gift registry link that included information about eythromelalgia and TEA.

It explained that EM afflicts some members of Todd's family. (Todd's stepsister Dr. Pam Costa and his stepmother Gayla Kanaster both have EM.)

And that one of TEA's goals is to fund research to find a cure. For instance, TEA has helped fund work at Yale School of Medicine and at Radboud University in The Netherlands.

They added the following statement:

"In lieu of a traditional gift, please consider making a donation to TEA as a wedding gift to us." TEA's Web site link followed.

One couple even held a pre-wedding party and asked guests for $20 donations.

Wedding gift donations totaled more than $2,700 in early Sept.

Besides not having to decide what to give, guests seemed pleased to be making a donation to a charity.

"Plus, how many times can you deduct a wedding gift from your taxes?" Todd asks.

Fundraising Challenge

(Continued from page 1)

Goals for future years include developing a full-blown campaign to inform doctors and other health care practitioners about EM, which will be an even more expensive project.

An eight-year-old, all-volunteer agency, TEA to this point has depended on its members to do the work of the organization.

Including raise money for it.

This summer, Todd and Karen Kanaster asked for donations to TEA in lieu of wedding gifts. (See story, above.)

In 2004, member Sarah Sundstrom gave hundreds of hours and organized an auction in Seattle, raising over $27,000 for research.

Other individuals have done their own fundraisers in their workplaces. Or had their grandchildren sell pizza discount cards.

If TEA is to grow and mature as an organization, raising funds for general use is a must.

So watch your mail for the appeal. And remember anyone can donate anytime on TEA's Web site.

Thank You Donors

TEA thanks the people and organizations who made donations to TEA in the six months from December 1, 2006, through May 31, 2007.

*gifts made in honor or in memory of someone
+gifts to the Research Fund

Zev Ancel+
Amy Bernardo+
Dolores Besch+
D. Cunningham+
Jessica Curtis+
David Dobbs+
W. Domenichelli+
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Networking Without Computers

Are you a member without a computer or computer skills? Or do you have to travel to your local library to use a computer?

If you answered "yes," you do not have easy access to TEA's Web site. So you can't use the member directory to contact others living near you who also have EM. Also, you can't read the medical journal articles or the "In the News" articles like "The Pain Gate" (Story on page 1.)

Then, just for you, TEA offers a service—the Networking Program. Co-chairpersons Gayla Kanaster or Judy Reese can give you the names of other people with EM who want to network. And they can make copies of articles and mail them to you. (There is a small charge for mailing.)

Just fill out the form in the adjacent column and send it to **Judy Reese, 1155 E. Wild Duck Lane, Salt Lake City, Utah, USA 84117.**

I want to participate in the TEA Networking Program.

Signature_____
Name (Please print)
_____

Street_____

City_____

Province_____

Country_____

Zip/Postal code_____

(Optional)
Phone_____
E-mail_____

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