

# FootSteps

The newsletter for members of The Erythromelalgia Association  
Footsteps online: [www.erythromelalgia.org](http://www.erythromelalgia.org) or [www.burningfeet.org](http://www.burningfeet.org)

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SPECIAL RESEARCH EDITION

## Why should TEA fund research at Yale?

Since 2004, TEA has donated most of its research dollars to studies at Yale that focus on the inherited form of EM. Most of TEA's members have EM that is not inherited, so why should TEA keep donating funds to Yale? TEA asked this question to EM researcher Stephen G. Waxman, M.D., Ph.D., Director of Yale's Center for Neuroscience and Regeneration Research, and got this response:

"Years ago, research on mutations from rare families with abnormal blood lipids pointed the way to development of lipid-lowering drugs (Lipitor) that not only help those families, but help millions of people around the world," Dr. Waxman wrote.

"Similarly, research on EM mutations is teaching us valuable lessons that will apply to many patients with pain, not just those families. We are learning from the mutations in people with inherited EM ... very valuable lessons about the critical molecular players in producing pain. By identifying those pain-producing molecules, we are helping the pharmaceutical industry target their efforts as they develop new and more effective pain medications."

"I would predict that these new medications, when available, should help all people with EM, with and without mutations, primary and secondary EM," Dr. Waxman wrote.

Yale researchers also believe drugs developed to block EM pain should help burning pain in general. What's causing the pain of inherited EM (an over-active molecular battery in pain-signaling nerve cells) does not "just produce pain in people with inherited EM who have mutations. There is good evidence that the same molecule is a key player in pain sensation in EVERYBODY, including people with EM (*continued on PG3*)

## And the Paint Your Pain winners are

After careful consideration, a panel of judges selected "EM" by Andrea Davenport as the first place finisher in TEA's Paint Your Pain artistic challenge. The painting features a figure standing on hot coals with flames licking up the legs. Both the hands and the entire face are also engulfed in flames. (*continued on PG4*)



The Erythromelalgia  
Association

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# FootSteps

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## The Erythromelalgia Association

[www.erythromelalgia.org](http://www.erythromelalgia.org)

200 Old Castle Lane,  
Wallingford, PA, USA 19086

Information/Memberservices  
(610) 566-0797

Email  
[memberservices@burningfeet.org](mailto:memberservices@burningfeet.org)

President  
**Beth Coimbra**

Vice President  
**Isabelle Davis**

Secretary  
**Deborah Mosarski**

Chair, Networking Program  
**Gayla Kanaster**

FootSteps Editor,  
Principal Writer  
**Isabelle Davis**

Contact  
[Isabelle.Davis@gmail.com](mailto:Isabelle.Davis@gmail.com)

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The intent and purpose of this publication is to inform those with EM or their friends and families—not, in any way, to provide medical advice.

## Research update: MAC members report status of EM research



Jay S. Cohen, M.D.

**Editor's Note:** Jay S. Cohen, M.D., Chairman, TEA Medical Advisory Committee, wrote this summary of his personal research into EM. To bring TEA members up to date on the status of EM treatment and research, he also asked other members of the committee to submit summaries of their work. See the report from the Mayo Clinic and Joost PH Drenth's summary of his research on PG7.

By Jay S. Cohen, M.D., Chairman, TEA Medical Advisory Committee

My research over the last 17 years has involved finding effective treatments to help me overcome five years of being bedridden and twelve years overall of disability from EM. In 1995, there was very little information about EM. None of my doctors had heard of it, nor had I. A podiatrist I met by accident made the diagnosis. I still have severe EM and without my treatments I would be bedridden within days. People have asked what my regimen contains. Here are my major therapies: Phosphatidylcholine (PC), a natural supplement available at [bodybio.com](http://bodybio.com) and at [millerpharmaceutical.com](http://millerpharmaceutical.com). (continued on PG7)

## TEA news briefs

### Membership renewals lag.

As of Sept. 1, TEA had received \$7,872 from members paying for new memberships and renewals at the suggested \$20 or larger donations. TEA thanks these mem-

bers for their donations. However, many past members still need to renew, which you can do easily on the website. If you don't use computers, please send your membership renewal donation to TEA, 200 Old Castle Lane, Wallingford, PA, USA 19086. TEA uses the membership donations to fund its \$17,000 annual operating budget. (Any amount of money you give when you renew is a tax deductible donation in the U.S.) Membership renewals (dues) are now due annually in June. You will not receive a letter telling you it's time to renew as in the past.

**Volunteer compiles resources.** TEA is now linked to the websites of other organizations representing disorders whose symptoms are similar or overlap EM's, thanks to the efforts of member Jackie Le of Orlando, Fla., U.S. Links are on TEA's website – go to "What is EM?" and click on "Links." Jackie is one of those who answered a request for volunteers and immediately agreed to take on the project. She sent letters to each organization, describing EM and TEA, asking them to add our link to their websites and offering to link theirs with TEA's. **Brochures available.** Extra copies of TEA's revised brochure are available for member's use. To request brochures, email [memberservices@burningfeet.org](mailto:memberservices@burningfeet.org).





## The Yale Research Team



## Why should TEA fund research at Yale? (continued from PG1)

without mutations," Dr. Waxman wrote.

TEA began supporting Yale's team of researchers in 2004 when Yale first made the connection between mutations to gene SCN9A and EM's burning pain.

Working with Yale, global pharmaceutical company Pfizer Inc. has a good candidate drug that could be starting clinical (in humans) trials in the "hopefully not too distant future." When the first trials take place, the company will want to focus on people with EM who have genetic mutations because of the scientific evidence their mutations cause EM pain.

"Mutations are leading the way ... much of the recent research is scientifically propelled by what we learn in people with EM due to mutations but the lessons from this research are highly relevant to chronic pain of all types," Dr. Waxman wrote.

Recent studies show what's causing the pain of inherited EM "is a major contributor to pain after nerve injury, as occurs after trauma, or as a result of nerve damage in diabetes. This sort of pain occurs in the general population and does not require a mutation ... it's also responsible for burning-type pain after burn injury and for burning pain in response to inflammation, for example, a bee sting." (See three new research studies the on the Articles page on the TEA website [www.burningfeet.org](http://www.burningfeet.org).)

"I am a physician and I know that people with EM are waiting for new and more effective treatments. I am very interested in alleviating pain in people with EM, and the lessons that we learn from mutations will hopefully get us there." Dr. Waxman concluded.

## Mayo study finds sensory abnormalities in EM patients

A number of sensory abnormalities were found in patients diagnosed with EM at the Mayo Clinic, a recent study concluded. Published this year in the British journal *Clinical and Experimental Dermatology*, the study lends support to the conclusion that neuropathy underlies the EM diagnosis.

Reviewing the medical records of 41 patients diagnosed with EM between 1994 and 2008 at the Rochester, Minn., U.S. facility, the Mayo team, under the direction of Mark D. P. Davis, M.D., looked at the results of computer-assisted sensory tests that had been among the evaluation measures used to determine the EM diagnosis. Included were quantitative tests for sensory responses to heat, vibration, and cooling.

Of the 41 patients, 82.9 percent had abnormal results, which confirms that the sensory status of most patients with EM is abnormal. The most common finding was a hypersensitivity to heat. This quantifi-

able sensory abnormality corresponds closely with the clinical finding of warmth, swelling and discomfort in the limbs. "These findings lend support to the notion that neuropathy underlies the EM diagnosis," the study concludes.

One factor that may bias these findings is the Mayo Clinic's status as a referral center that receives the most severely affected patients who have seen many other physicians and may have travelled many miles for evaluation. Thus the patients in this study may not be representative of the general population of EM patients, the study cautioned.



Mark D. P. Davis, M.D.

# And the winners are



"EM" by Andrea Davenport  
1st Prize

“ The judges had to pick winners, but every one of the submissions is a gift to TEA and its members ”

Deborah Mosarski  
TEA secretary

(continued from PG1) Suggested by researchers at Yale, the artistic challenge began with an announcement on TEA's website and in the Spring 2012 issue of *FootSteps*. The 17 entries submitted are "both moving and emotionally difficult to look at," judges told challenge organizer and TEA Secretary Deborah Mosarski.

Judges also told her seeing the paintings helped them understand what EM was more than reading an article ever could. "The judges had to pick winners, but every one of the submissions is a gift to TEA and its members," said Deborah.

Second prize went to "A Constant Battle" by Jennifer Beech, a contemporary painting of large hands bringing heat to the face and feet of a screaming individual.

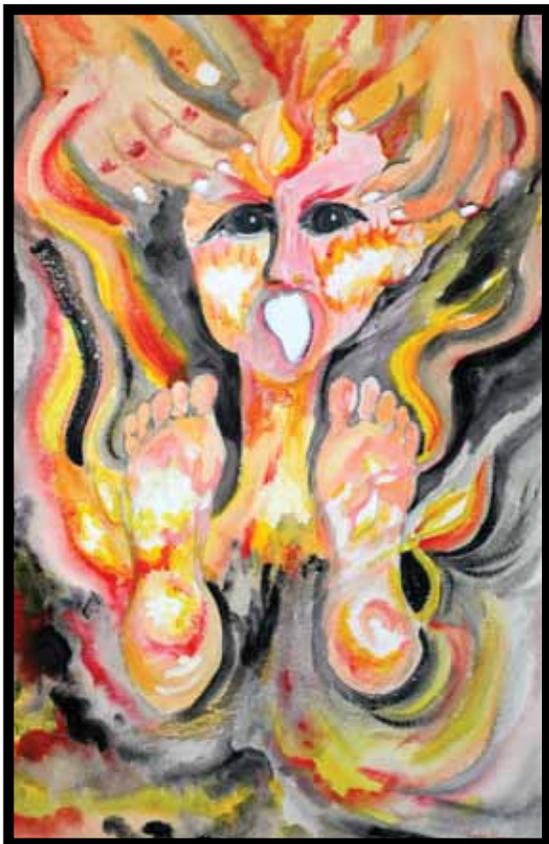
Heidi Grein's "Red Heat" tied for third place. Heidi describes her painting as "An electric shock seems to have hit your normally functioning body parts. 'Red Heat' spreads out, a thunderstorm of pain." Judged as equally as impactful is "Red Thief" by Audrey Bell.

Honorable Mention is a touching depiction of a young girl whose feet are bound in a firepit, titled "Chained to Fire" by Bailey Deacon. "This girl is stuck in pain and no matter how hard she tries to escape the pain she can't," Bailey wrote.

The "winnings" TEA awarded were complimentary memberships to TEA: first place—five years, second place—three years, third place and Honorable Mention—two years, and to show TEA's appreciation for participating, all entrants received one year's membership.

The winners along with the other paintings/drawings are on TEA's website [www.erythromelalgia.org](http://www.erythromelalgia.org).

# Artistic challenge results



"A Constant Battle" by Jennifer Beech  
2nd Prize



"Red Heat" by Heidi Grein  
3rd Prize - Tied



"Chained to Fire" by Bailey Deacon  
Honorable Mention



"Red Thief" by Audrey Bell  
3rd Prize - Tied

# TEA recognizes 2011 donors - thank you!

Thanks to these people and companies for making donations to TEA in 2011.

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## Drenth's research team searches for genetic variants

By Joost PH Drenth, M.D., PhD., TEA Medical Advisory Committee member

Researchers in the Radboud University Nijmegen Medical Center in Nijmegen, The Netherlands, are providing genetic testing for erythromelalgia (erythromelalgia). We knew for many years that EM can be inherited within families. A few years ago the gene for EM was discovered by advanced molecular techniques. This gene encodes a sodium channel that is located on nerve endings. In some patients with EM there are variants within this gene that cause the disease.

With the same techniques we screen many patients who have symptoms reminiscent of EM. Over the years we have discovered that genetic variants within this gene can cause a spectrum of diseases. Indeed, some variants cause a syndrome featured by extreme pain, while other variants that impair the function of the gene cause patients to not feel pain at all. Recently we

and others have picked up variants that have been associated with subtle changes in the small nerve endings that cause pain.

These findings were only possible because we are able to screen patients with various pain pathologies for these genetic variants. Our laboratory has screened 450 patients.

In some 18 percent we have found bona fide changes in the gene. This means that there are more genes out there that can cause painful feet and hands. We are constantly searching in order to find new variants in the gene that we know and are looking for the genes that we do not know. The search continues.



Joost PH Drenth,  
M.D., PhD

## EM patients get thorough assessment at Mayo

TEA Medical Advisory Committee member Mark D. P. Davis, M.D., summarized the Mayo Clinic's work in EM: "We continue to see and assess patients with erythromelalgia at Mayo Clinic. Patients have a detailed assessment when here which generally includes blood tests, vascular and neurologic studies and assessment.

Comprehensive treatment strategies are available and individualized to the patient, with treatment approaches ranging from topical medications to systemic medications to interventions for chronic pain, as appropriate."



## Research update: MAC Chairman reports *(continued from PG2)*

Other types of PC, which are based on soy, do not work. Alpha lipoic acid (ALA), a natural supplement known to benefit neuropathies: use up to 1200 mg per day in divided doses; avoid "R-ALA." Another natural therapy worth trying is black cohosh, an herb used for hot flashes in menopausal women.

What is the therapy that helps me the most? Sandomigran (pizotifen), an antihistamine and anti-serotonin drug from Canada. Similar to cyproheptadine (Periactin), which helped me slightly, Sandomigran is the best substance I have ever taken for my EM. There are two reports in the medical journals of its benefiting people with EM including some members of a family with familial EM.

None of these or other treatments will help everyone with EM. Although our disorders may look similar, the underlying physiology varies greatly from person to person. **My secret of regaining a good life is to keep trying things, doing so carefully, gradually, one at a time, and stopping any substance that makes your EM worse, while sticking with the ones that help.** For many of us, no one drug will adequately control EM. Instead, a combination of therapies may be needed. Also, because there are so few doctors who know how to treat EM, just find a doctor who is interested in you and willing to follow the guidelines available here at TEA and at my website, MedicationSense.com.



The Erythromelalgia Association

# FootSteps

**Raising Awareness.**  
**Raising Research Funds.**  
**Raising Hope.**

[www.erythromelalgia.org](http://www.erythromelalgia.org)

## **Text to donate: a new way to raise funds**

TEA members raised \$560 in \$10 donations one month this summer by sending text message donations to TEA. Those gifts were matched 100 percent by the EveryLife Foundation, raising the total dollars to \$1,120. The EveryLife Foundation offered this "rare opportunity" for matching funds to rare disease organizations like TEA that have partnered with its campaign to "Cure the Process" for developing treatments, especially drugs, for rare or "orphan" diseases.

EveryLife gave TEA members from July 19 through August 18, 2012 to use a "Text to Donate" feature. Donations had to be texted from donor cell phones to TEA's website \$10 at a time. TEA sent an email to members to alert them of the opportunity. TEA member Kelly Ballard, Jacksonville, Fla., U.S., approached her co-workers and family and raised \$240 of the \$560 in text donations herself.

Another TEA member has agreed to investigate placing a permanent text-to-donate feature on TEA's website that would allow donation texts to be sent from anywhere in the world.



200 Old Castle Lane,  
Wallingford, PA, USA 19086

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