FootSteps

The newsletter for members of The Erythromelalgia Association
FootSteps online: erythromelalgia.org or burningfeet.org

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TEA gives $75,000 to Yale research

Photo by Daria Sizova, Ph.D.

TEA's Board of Directors voted this summer to donate $75,000 to Yale's Neuroscience and Neurorehabilitation Research Center for studies into inherited EM and related pain disorders. At the discretion of the center's director Stephen G. Waxman, M.D., Ph.D., the money will pay directly for costs like salaries, supplies and equipment. "Private dollars (like those from TEA) give us stability, allow us to move more rapidly, and give us the flexibility to try new things...." said Dr. Waxman, Yale professor of Neurology, Neurobiology and Pharmacology.

Since proving that a single genetic mutation (in sodium channel Nav1.7) causes the pain of inherited EM, scientists at this large, multidisciplinary laboratory—now 30 strong—have used IEM as a human genetic model of neuropathic (nerve) pain. Through the use of twenty-first century technology, these scientists are unlocking secrets about how our bodies send pain signals to our brains from many sources, not just the skin as in EM.

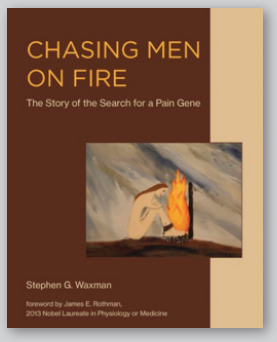
The most recent Yale study that TEA has on its website appeared in the *2016 JAMA Neurology*. It reported that using new technology, it is possible to guide drug treatment by using genetic information and molecular modeling. An accompanying editorial declared, "There are still relatively few examples in medicine where molecular reasoning is rewarded with a comparable degree of success...."

TEA made its first donation ($60,000) to Yale in 2004. Before that TEA helped the scientists collect blood samples from members with inherited EM and their family members. Analyzing blood cells showed genetic mutations that made pain-sensing nerve cells hyper-excitable, firing off pain signals when they should not. Next the researchers proved that these mutations caused the pain of inherited EM. Study results were published in 2005. (See Medical Articles on TEA's website erythromelalgia.org.)

Waxman book published

Chasing Men on Fire, The Story of the Search for a Pain Gene by Stephen G. Waxman, M.D., Ph.D., was published in 2018. The book tells the story of a thirty-year scientific journey that spans three continents and involves the DNA of people with a rare genetic disorder —inherited erythromelalgia—who feel they are on fire. The book is described in the foreword by Nobel Laureate James Rothman as "so well written it reads like a detective novel." On the cover and throughout the book is artwork produced for TEA's 2012 Paint Your Pain contest. Dr. Waxman ends the book by predicting that the search will ultimately result in new medications, without side effects, that will treat the pain of people with man on fire syndrome and others.

Available at Barnes & Noble, Walmart, and Amazon.com.



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A Day with Beth

By Isabelle Davis

Rarely, if ever, will you find members of TEA's Board of Directors in the same place at the same time. That's because each of us lives in a different state in the USA. We communicate primarily by email and hold our monthly meetings by teleconference. In July, however, I had the great pleasure of spending a Saturday with Beth Coimbra, president of TEA. My husband planned a last minute trip to Wilmington, DE, which is within a half hour of Beth's home. I went with him and had the chance to get some "face time" with Beth.

Beth and I had met once before in March 2005. After TEA made its first donation to the Center for Neuroscience and Regeneration Research at Yale University, researchers invited representatives of TEA to visit the labs where research into inherited EM was under way. Then TEA vice president Beth and I decided to accept the invitation.

Beth and I made an instant connection back then, the sort of connection one only makes with someone else who has experienced the rare, puzzling, painful symptoms of EM. We shared our stories, but were mostly involved in our fascinating visit to the Yale research laboratory. This time we had lots of time to talk in person as Beth drove me to the Philadelphia, PA, suburb where I grew up. We went past the schools I attended (long ago) and then down the street where I lived and past the house where I grew up. She then hosted a delicious lunch at her home. We built a friendship that day, the sort you can not make by email and one I hope will last a long time.

Your stories: everyone has one

Carolyn Quinn
Plumas Lake, CA, USA

My story begins in 2002 when I broke my fifth metatarsal (the last long bone on the outside of the foot). After a few months of (supposed) healing, my feet began to burn and turn red. I googled "hot feet" and came up with Erythromelalgia. The EM symptoms have since spread to my hands, but are not as severe there. I have also had Raynaud's syndrome for many years. Since developing EM, I've tried many different meds, but presently I'm taking: Cymbalta, Celebrex and Trazadone (for sleep). These three meds have given me pain relief and increased the length of time I can walk and be in the sun. However, no matter what activity I want to do, I have to ask: "Can I handle this with EM"? Others in my life (except for my husband and BFF) seem to forget that I have this condition, as I don't complain. Still, EM has changed my life drastically. I'm now 67 and I'm praying for " the cure" to come soon! Swimming has been my salvation!

MAYO CLINIC

Study finds certain EM patients respond well to corticosteroids

As reported in the *Journal of the American Academy of Dermatology*, physicians at the Mayo Clinic have identified several factors that may predict an EM patient's responsiveness to corticosteroid treatment. In a retrospective study, a search of clinic records from 2000 to 2011 identified 31 EM patients who had received corticosteroid treatment (71% were female, and the median age was 47 years). The study reports that 55% of those treated saw improvement and categorized this group as "steroid responders." Among the responders were 29% complete responders, who had minimal to no EM pain following treatment and 26% partial responders, who had mild to moderate persistence of symptoms. The other forty-five percent of patients treated saw no improvement and were classified as steroid nonresponders.

> Of the patients who reached maximum pain intensity within 21 days of onset, 87% responded to steroids.

Several clear trends were discovered. The first factor investigated was how quickly the patients' EM symptoms peaked. Of the patients who reached maximum pain intensity within 21 days of onset, 87% responded to steroids. The study's principal investigator, Gabriel Pagani-Estévez, M.D., says that this rapid onset suggests a "prominent inflammatory component" to those patients' EM, and it stands to reason that corticosteroids, a class of anti-inflammatory drugs, would prove effective. Furthering his theory, of the patients who were complete steroid responders, 67% reported a disease trigger such as surgery, trauma, or infection. (It should be noted, however, that these events have not been definitively proven to have caused EM in any of these cases, only that their timing precipitated the onset of symptoms.) Another factor studied was how promptly the patients were treated with steroids following diagnosis.

Researchers found that steroid responders had been diagnosed with EM earlier than nonresponders, at an average of 6 months vs. 24 months following the onset of symptoms. They also received steroid treatment earlier, at an average of 3 months vs. 24 months following onset.

> ...a "golden window" may exist in some patients, a period where steroid intervention is most useful.

Finally, a high dose of corticosteroids tended to be more effective than a lower dose. The majority of patients who saw complete elimination of their symptoms were on the highest dose regimen of 1,000 mg of IV methylprednisolone per day. Of those patients who received a high oral prednisone dose of at least 40 mg administered daily for a minimum of five days, most (76%) had either a partial improvement or total improvement (but a smaller percentage of these patients had complete relief than did those who received the very high dose IV steroid). Dr. Pagani-Estévez believes more patients might have responded had their steroid dose been higher. He also believes that a "golden window" may exist in some patients, a period where steroid intervention is most useful. He says, if not treated promptly enough, irreversible damage to peripheral nociceptors and central sensitization may occur, making an initially reversible process irreversible. Dr. Pagani-Estévez's concluding advice to physicians: "If you are going to do a steroid trial, hit these patients hard with steroids and hit them early."

Dr. Gabriel Pagani-Estévez, M.D. is a Mayo Clinic trained, board certified neurologist with solid clinical and extensive research experience.

The Erythromelalgia Association

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TEA encourages you to share your story about how EM has affected your life. Please consider sending a "head shot" and your story (350 words or less) to **memberservices@burningfeet.org** or mail to **200 Old Castle Lane, Wallingford, PA,19086**.

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State proclaims EM Awareness Month

May 2018 was EM Awareness Month in Massachusetts, USA, by proclamation of the governor of the state. On May 9, 2018, a Massachusetts state representative presented the proclamation in front of approximately 40 people at the State House in Boston, MA. TEA member and Massachusetts resident Paula Corey, who lives with EM, organized this awareness event that gathered people with EM, their friends and family. Among the guest speakers addressing the group was Anne Oaklander, M.D., Massachusetts General Hospital, and TEA President Beth Coimbra.

The proclamation described EM as "intense burning pain of the affected extremities" and that "the cause of EM remains unknown in the vast majority of cases". The document further explained that people with EM must often make "major adjustments to their lifestyles".