

ON THE GO

Essential Tremor hasn't stopped Stephen Remillard from competing at a very high level, athletically and professionally.



Essential Facts about Essential Tremor

This “quiet” disease, which affects 10 million Americans, is anything but benign.

BY STEPHANIE STEPHENS

The questions from strangers roll right off Stephen Remillard's back—questions like “Why are you so nervous?” and “Don't you think you'd better ease up on the caffeine?” At age 26, Remillard has faced down essential tremor (ET)—and these sometimes obnoxious personal inquiries—“for as long as I can remember,” he says.

Diagnosed at age five, Remillard is one of an estimated 10 million people in the United States with ET, according to the International Essential Tremor Foundation (IETF). Never heard of essential tremor? Perhaps it's because so many other conditions compete for headlines. Advocates for people with ET have not had the benefit of a celebrity spokesperson such as Michael J. Fox or Muhammad Ali, who both have Parkinson's disease. The most famous person with ET was the late actress Katherine Hepburn, who parlayed her disorder into an intriguing component of both her vocal and on-screen personae. Other notables with ET include retired Supreme

Court Justice Sandra Day O'Connor and the late West Virginia Senator Robert Byrd, who passed away last summer.

Although the disease is not fatal, it can cause severe disability. Even when symptoms are mild, people with ET often feel frustrated or embarrassed by their condition, says Catherine S. Rice, M.N.M. (Master's in Nonprofit Management), executive director of the IETF. (See box, “The Faces of ET,” for a list of advocacy groups.)

WHAT IS ESSENTIAL TREMOR?

Essential tremor is a progressive neurologic condition that causes involuntary rhythmic trembling of the hands, head, tongue, voice, and less often the legs or trunk. Like Parkinson's disease, ET is considered a movement disorder. The disease is caused by abnormal communication—sometimes called a “misfire”—between certain areas of the brain including the cerebellum, thalamus, and brain stem.

“In many cases the tremor **will get worse**. You may lull yourself into thinking, ‘It isn’t life-threatening,’ but I know people who can’t write, feed, bathe, or dress themselves.”

—CATHERINE RICE, EXECUTIVE DIRECTOR, IETF

It is often hereditary, according to Kelly E. Lyons, Ph.D., president of the IETF and director of research and education at the Parkinson’s Disease and Movement Disorder Center at the University of Kansas Medical Center in Kansas City, KS. In fact, ET is also called hereditary or familial tremor. However, no one in Remillard’s family has had ET, to his knowledge. (See box, “Essential Tremor: The Basics.”)

Tremors may be more prominent when the person is highly active or anxious. A vicious circle can occur with ET: The person fears embarrassment in public, which in turn makes the tremor worse. As a result, people with ET may limit social interaction and even develop social phobia. Sometimes, observers assume the tremor is due to drug or alcohol intoxication.

“Essential tremor can cause significant disability,” says Dr. Lyons, who is not fond of the moniker “benign” sometimes added to the disease, as in “benign essential tremor.” “That label inappropriately minimizes its effects on many of those who have ET,” she says.

In addition, tremor tends to worsen with age. True to form, Remillard’s disorder—which began in his hands, progressed to his legs, and eventually affected his voice—became more recognizable as he matured, prompting those wise-cracking questions about caffeine. But the joke is definitely not on him. This energetic property-leasing consultant from the Los Angeles area is always on the run—literally. Remillard just finished his ninth big race, the Nov. 14 Malibu International Half Marathon: 13.1 miles in two hours and—so close!—one minute.

“The ocean-side, cliff-top path was more difficult than I’d ever imagined,” he says. Nevertheless, ET clearly hasn’t stopped Remil-

lard from competing at a very high level, athletically and professionally. “I’ve lived with ET and I’m able to ‘control’ it,” he says, even without medication. “I’m a pretty confident person, always seeking ways to overcome limitations. I make do with most situations I’m presented with, and I’m able to do anything anyone else can do.”

NOT THAT BAD—OR IS IT?

Remillard takes pride in being neither a complainer nor a whiner. But health care professionals actually wish more people with ET would speak up about their condition.

“It’s clear from studies that the majority of patients have not been to a doctor or diagnosed,” says Rodger J. Elble, M.D., Ph.D., director of the Parkinson’s Disease and Movement Disorders Center at Southern Illinois University School of Medicine in Springfield and an IETF medical advisory board member. The reason may be that the condition is initially mild—in fact, ET may remain mild for decades—or because a family member has ET and relatives become used to it, Dr. Elble says.

“Often as patients approach retirement or late middle age, the tremors become more noticeable,” he says. “Other people may develop tremor late in life—after age 65.”

Many in this age group may have ET and just not know it—they simply think tremors are due to age. Statistics show that in the general population, occurrence of ET is close to 1 percent, says Dr. Elble. “But if you analyze people 65 and older, the number is 5 percent.”

“Unless people are appropriately diagnosed we can’t help them,” says Rice. “In many cases the tremor is progressive and will get worse. You may lull yourself into thinking, ‘It doesn’t bother me and isn’t life-threatening,’ but I know 10 more people who can’t write, feed, bathe, or dress themselves. Their quality of life is severely impacted.” (For help locating a neurologist, use the AAN’s Find a Neurologist Tool at aan.com/patients.)

COMMONLY MISDIAGNOSED

Essential tremor can be easily misdiagnosed as another movement disorder, most frequently Parkinson’s disease. Essential tremor actually occurs eight times more often than Parkinson’s. A handwriting test is often used to distinguish between the two, says Dr. Lyons. “Essential tremor often results in large, shaky handwriting samples. Parkinson’s writing typically starts out normal, then gets smaller and smaller.”

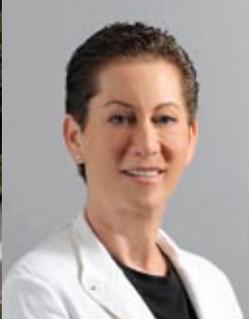
Tremors of the voice and head also do not generally occur in Parkinson’s but are commonly seen with ET. A patient suspected of ET should be examined while in a reclined or seated position with complete body support.

Dystonic tremor is also often confused with ET. Patients with

Essential Tremor: The Basics

- ▶ Essential tremor (ET) is a neurological movement disorder that causes the hands, head and voice, and sometimes the legs and trunk to shake.
- ▶ Approximately 10 million Americans have ET.
- ▶ Each child of a parent with the ET has a 50 percent chance of inheriting a gene that causes the disease, though people with no family history of tremor may also develop ET.
- ▶ Parkinson’s disease and dystonia are the most common misdiagnoses for ET.
- ▶ Essential tremor is frustrating, embarrassing, and at worst, disabling.





The Faces of ET

Essential tremor affects people from all walks of life and is not confined to the elderly. Children, adolescents, and young- and middle-aged adults can also have ET. Even newborns have been diagnosed with the condition.

The International Essential Tremor Foundation sends a free information packet about ET upon request, maintains a listing of physicians specializing in ET, and assists with forming ET support groups. Members receive two issues per year of the member magazine, *Tremor Talk*.

ESSENTIAL RESOURCES

- ▶ International Essential Tremor Foundation: essentialtremor.org
- ▶ Movement Disorder Society: movementdisorders.org
- ▶ WE MOVE: wemove.org

says Dr. Lyons. “If tremor is not sufficiently controlled with one of these medications, the two can be used in combination. If these medications do not control tremor or cause intolerable side effects, then gabapentin or topiramate are probably the next most commonly tried medications. Benzodiazepines are also used, especially if anxiety is also an issue, but the potential for addiction should be considered.”

According to IETF, new medications being tested for effectiveness and safety in the treatment of ET include 1-octanol, sodium oxybate, T2000, and carisbamate. (See Resource Central, p. 43.)

In addition to medication, people utilize a combination of creativity and tenacity to cope with ET. Using the tremor-free hand helps with completing tasks, as does holding one’s chin to the chest to control head tremor. Stress reduction also plays a vital role. (See the extensive list of “Coping Tips” on the IETF Web site at essentialtremor.org.)

Meanwhile, current research focuses on finding the gene that causes ET. In many large families, the condition appears to be dominantly inherited—that is, there’s a 50-50 chance each child has it. In research reported in April 2010, one particular gene (the LINGO1 gene) was found to be associated with increased risk of ET.

However, “LINGO1 is not a ‘Mendelian dominant trait,’ in which inheritance of the gene determines disease,” says Dr. Elble. In fact, LINGO1 is only associated with an increased risk of 1.3 to 1.5. Finding a dominantly inherited gene is viewed by experts as crucial to finding a cure.

“We do not have a shortage of interested researchers out there,” adds Catherine Rice, of the IETF. “But there aren’t enough researchers looking *only* at ET. We want to fund the research to help find the cause of essential tremor.”

IT IS BRAIN SURGERY

“I turn myself off when I go to bed and back on every morning,” says 60-year-old Shari Finsilver of Orchard Lake, MI. She manages ET with a “brain pacemaker” device that delivers

dystonia exhibit an abnormal, involuntary spasm or contraction of muscles in the face, neck, hand, or torso. When muscles aren’t used, they normally relax. With dystonia they do not, resulting in abnormal postures.

General neurologists treating tremor have a diagnostic error rate of 25 percent to 35 percent. “There is no laboratory test for ET, no biological marker to diagnose it,” Dr. Elble says. “Diagnosis is made by careful history and exam.”

MEDICATIONS AND RESEARCH

One New York-based study showed only 8 percent of patients had actually been prescribed medication for tremor. Furthermore, patients who try one or more medications for treatment usually find them to be less than satisfactory. No medication has been specifically developed to treat ET.

“If a good, relatively inexpensive, and safe treatment were available, a lot more people would take it,” says Dr. Elble.

For approximately 60 percent of patients, medications are available that might help alone or in combination, although “some patients will not respond at all,” says Dr. Elble.

Experts consider the best medications to be propranolol, an FDA-approved beta blocker, and primidone, an anti-epileptic, according to Dr. Elble. The anti-anxiety medications clonazepam, diazepam, lorazepam, and alprazolam, and the anti-convulsants gabapentin and topiramate, have also shown some benefit for ET.

“Most experts agree that primidone is the most effective treatment for ET, followed by propranolol and other beta-blockers,”

PHOTOS COURTESY OF THE INTERNATIONAL ESSENTIAL TREMOR FOUNDATION

Medication and/or **deep brain stimulation** can help some people with essential tremor.

electrical stimulation to her brain. The therapy is known as deep brain stimulation (DBS).

“In general, essential tremor is the simplest of diseases we treat with DBS,” says Joseph Neimat, M.D., M.S., neurosurgeon at Vanderbilt University in Nashville, TN.

“It is a single, prominent symptom that responds very well to intervention. We can see effects almost immediately—and it is probably effective in 90 percent of patients.” [Disclosure: Dr. Neimat says, “I have done some very limited consulting for Medtronic, a manufacturer of the device, for a few hundred dollars. I have also done a small amount of teaching for Medtronic and another company that makes frames for the device—again, for a small fee.”]

Finsilver is a past president of the IETF and current executive board member, and she has become a spokesperson for seeking diagnosis and treatment of ET. Considering herself lucky to have been diagnosed immediately by her neurologist at age 19, Finsilver says the procedure “has been the life-changing solution that medication could not provide.”

In her 40s, she’d had enough of her tremors and of medication side effects. “With one drug, I couldn’t lift my head up off the pillow.”

Finsilver couldn’t write, draw, or apply eye-makeup due to her uncontrollably shaking hands. One wonderfully fateful day in 1996, her sister called, ecstatic about a TV news report she’d seen on DBS.

The concept of brain surgery doesn’t resonate with everyone, though. “There are hurdles every patient has to get past,” says Dr. Neimat. “Many people have an internal prohibition to having someone open up their head.”

Finsilver took her time to learn about DBS and finally underwent the procedure at Henry Ford Hospital in Detroit, MI, in February 1999. Before doing so, this grandmother of five with a master’s degree in industrial and operations engineering taught herself relaxation. She experienced few problems, even with the notorious “halo.” This metal frame immobilizes the head with four screws and allows mapping of the brain “target.”

With use of only topical anesthetic, holes are drilled into the head and an electrode is placed in the brain. Wires running beneath the scalp connect to a pulse generator—this is the “brain pacemaker”—underneath the skin of the upper chest. General anesthesia is used for the last phase of the surgery.

Since 1995, more than 80,000 patients have received DBS. As a result of surgery, certain types of hemorrhage may occur in 1 to 5 percent of patients, seizure in 3.1 percent, and confusion during the recovery period in 1 to 36 percent, according to the new book, *Deep Brain Stimulation Management*,

edited by William Marks Jr., (Cambridge University Press, 2011). Among device-related problems, infection may occur in 3 to 10 percent; skin erosion in 2.5 to 6.45 percent; and fractures, migration, or malfunction of device parts in 5.1 to 18 percent.

The procedure itself is about “location, location, location,” explains Dr. Neimat—specifically, targeting the right spot in the thalamus. This delicate substructure of the brain relays sensory information to the cerebral cortex, which is responsible for processing information and for language. The specific area of the thalamus targeted for ET is densely packed against other important areas that provide sensation and carry motor fibers. Inadvertent stimulation of these other areas may produce their own effects in the body. For example, a patient might experience tingling when the system is turned on or muscle contractions if turned up too high. If an electrode isn’t in exactly the right location, speech problems can occur.

Following insertion, both physician and patient must make friends with the device, which requires “tweaking” to optimally adjust stimulation. This getting-to-know-you period requires patience from both parties.

Still, the future holds great promise for the technology, says Dr. Neimat. Already, newer systems feature rechargeable batteries that last longer, allowing for more patient-administered preprogramming. “Smart machines” will eventually measure the tremor and adjust maximally to reduce it, he says.

Deep brain stimulation is approved in this country for treatment of ET, Parkinson’s disease, dystonia, and obsessive-compulsive disorder. Ongoing studies here and abroad are testing DBS for severe depression, anorexia, severe epilepsy, Tourette syndrome, obesity, and early Alzheimer’s disease.

Finsilver, of contagious good spirit, is not afraid to share what she originally kept hush-hush. She formed an in-person and online tremor support group in 2001; in doing so, she realized that for years she had hidden both herself and her disease. Although she’s moved on from the group, she thanks those friends she made who encouraged her to proclaim: “Stuff it! This is who I am.”

She has few regrets. “I’ve always said I’d do DBS again,” Finsilver says, admitting that what’s done isn’t always finished. She’s endured having to replace “a wire with bad insulation that shot electricity through my body.” And recently a different wire broke. Batteries must be changed every three to five years, requiring minor incision under local anesthesia.

Those patients who thrive after DBS know that almost everything requires a little maintenance to keep it working optimally—even a brain. NN