



Essential Tremor (ET)

The driving need for ET research

Ludy Shih, MD

Essential tremor is a common but misunderstood disorder affecting the lives of millions of Americans. As a movement disorders neurologist, I see this firsthand in the patients that come to clinic looking for help in treating their tremor. Patients are affected in different ways and respond to treatments in varying degrees. Overall, tremor and tremor-related disability tend to get worse with time.

As a clinician investigator, I see two things driving the need for essential tremor research: A) the need for more effective and better tolerated medications for tremor, and B) the ways that the common clinical features of ET should lend us clues to the understanding of tremor.

To illuminate on point A: Tremor medications that are currently used are a byproduct of research and development on other neurological conditions like epilepsy. Because little is known about how tremor works in humans and how it might be manipulated successfully, pharmaceutical companies have little incentive to develop a robust product development program for tremor.

To expand on point B: Many ET cases are familial, providing hope that genetic mutations can be identified that will reveal basic knowledge about how tremor works in humans, similar to what the discovery of genes causing Parkinson's disease has done for that field.

If we are fortunate, genetic mutations may reveal a chemical target that can be manipulated with the right drug, or they may reveal a more complex mechanism rooted in nervous system development or even neurodegeneration although this latter point is quite controversial. Our research at Beth Israel Deaconess Medical Center focuses on two areas.

One goal is to try to identify patients who have subtle structural brain changes that may be associated with a more rapid progression of tremor related disability. Also, we are trying to identify whether there is any association with genetic variations in a gene called LINGO1 that is not seriously defective in people with ET but may contain slight variations that help contribute

to the development of essential tremor in a patient.

The second goal is to work collaboratively with other centers through the North American Essential Tremor Consortium to try to build a robust, data-rich repository on a large group of patients in order to classify and identify biological markers of tremor that might help enrich our genetic studies. Genetic techniques have advanced significantly over the years but there are many forms of tremor. Each may have their own mechanism and methods of treatment. Therefore, careful clinical characterization may still be necessary in order to make the proverbial "finding of a needle in a haystack" more productive.

Patients can help by volunteering to be a part of these types of studies and by raising awareness in their community about the need for essential tremor research. Like any difficult endeavor, persistence, teamwork, collaboration and organization among patients, their advocates, physicians, and scientists are often the key to success.

Continued support for the IETF—from people like you—is crucial to sustaining our research efforts. "Your donations have made a significant difference in this research," says IETF Executive Director Catherine Rice. "The IETF has provided funding for genetic studies in the past and continues to fund critical research in this area. We thank you for your continued support."



The mission of the International Essential Tremor Foundation is to provide hope to the essential tremor (ET) community worldwide through awareness, education, support and research.