science café

Epilepsy – in Search of Genomes with Frustrating Phenomes: A Conversation with Dan Lowenstein

By Jeffrey Norris

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Dan Lowenstein

Epilepsy was familiar to the ancients, including the Greek physician Hippocrates. But it wasn't until the last century that EEG recordings began to offer more insight into the brain's electrical activity during a seizure. And it is only in the most recent decades that researchers have begun to make more significant strides in understanding epilepsy – in terms of anatomical abnormalities, genetic mutations, and proteins called ion channels — and their effects on how electrical signals are transmitted within groupings of nerve cells in the brain.

Still, the causes of the most common forms of epilepsy remain elusive. Now, in what he calls the biggest epilepsy research project in history, UCSF neurologist Dan Lowenstein, MD, along with colleagues at 13 major epilepsy centers across the country, is looking for genes that are associated with common – and some less common – forms of the disease.

The $15 million clinical study, called the Epilepsy Phenome/Genome Project, is recruiting about 3,000 patients. For the most common forms of epilepsy, the researchers are looking for families in which at least two siblings are affected.

If the researchers identify genetic variations or mutations that can be implicated in the types of epilepsy they are studying, it will trigger more research on what those genes and their associated proteins do to contribute to specific forms of disease. That, in turn, is expected to lead to better insight into which drugs ought to work best for particular patients. Success in the project may also lead to the design of entirely new drugs.

We learn more about genomes, phenomes and epilepsy this week on the UCSF Science Café.

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