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UCSF-Led Team Receives \$15 Million To Study Genetics Of Epilepsy

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A team led by UCSF scientists has received a grant of \$15 million, provided over five years, to study the complex genetic factors that underlie some of the most common forms of epilepsy.

The study, known as the "Epilepsy Phenome/Genome Project (EPGP)", is funded by the National Institute of Neurological Disorders and Stroke, and brings together more than 50 researchers and clinicians from 15 medical centers around the country.

The project is led by Daniel Lowenstein, MD, professor and vice chairman of the Department of Neurology at the University of California, San Francisco (UCSF), and director of the UCSF Epilepsy Center. Lowenstein and Ruben Kuzniecky, professor and director of research in the Department of Neurology at the New York University Comprehensive Epilepsy Center, are co-principle investigators of the project.

Epilepsy is among the most common neurological disorders in the world, affecting one in every 100-200 people. It is characterized by seizures caused by abnormal electrical activity between nerve cells in the brain.

Although it has been known since antiquity that the disorder is influenced by inherited genes, progress to date has been limited to the discovery of single gene mutations that cause the disease in a relatively few number of families, says Lowenstein. For the more common types of epilepsy, heredity plays a more subtle role, and it is thought that a combination of variations in multiple genes likely determine an individual's susceptibility to the disorder, as well as the responsiveness to anti-epileptic medications.

In their study, the scientists will work to identify the constellation of genes that contribute to the more common types of epilepsy. The long term goal of the project, says Lowenstein, will be to identify potential molecular targets that could be the basis of much more specific and effective treatments for patients who have epilepsy, and the prevention of epilepsy in those at risk.

Because the approach to teasing apart the more complicated genetic factors in epilepsy requires a very large number of patients whose epilepsy has been extremely well-characterized, the investigators will enroll 3,750 patients and 3,000 people who do not have the disorder - so-called "controls" used for comparison -- over the course of the study.

Details about each patient's disorder - type of seizures, results of electroencephalograms and imaging studies, and effects of treatment -- will be collected and archived in a central data repository at UCSF, and all participants will be asked to submit a sample of blood or saliva as a source of their DNA. (All the clinical information and the DNA samples will be de-identified so that it cannot be traced back to a specific individual.)

Once this first phase of the study is completed, Neil Risch, PhD, and colleagues in the UCSF Institute for Human Genetics, along with researchers at Emory University, will carry out "whole genome scans" and look for potential connections between patterns of DNA sequences and specific characteristics of epilepsy in the study population.

"This is an extremely exciting step in the effort to illuminate the underlying causes of the common forms of epilepsy," says Risch, UCSF Lamond Distinguished Professor in Human Genetics. "The endeavor illustrates the type of research now taking place in human genetics thanks to advances in technology and our understanding of the human genome."

Additional information on the EPGP, including enrollment in the study, can be found at www.epgp.org.

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