



## **FAMILY STUDIES IN EPILEPSY RESEARCH**

### *Information Sheet*

**THE FAMILY STUDIES IN EPILEPSY PROGRAM** is a research program to discover genes that influence adolescent or childhood Idiopathic Generalized Epilepsy (IGE). Directed by David A. Greenberg, PhD, the study is located in the Mailman School of Public Health, Columbia University in New York City.

Epilepsy affects about 1% of the population (2.5 million) in the U.S and is classified as a complex common disease, a mixture of conditions that often have the same symptoms and characteristics, but have different causes. Nearly 50% of all epilepsy cases are of unknown origin – they seem to “just happen.” These epilepsies are termed “idiopathic.” Some of these idiopathic epilepsies are Generalized Epilepsies because they do not start from a specific spot in the brain. Such epilepsies are known to have a strong genetic basis.

**THE STUDY TEAM:** Our research is designed to identify genes that either cause epilepsy or make it likely one can develop epilepsy. Such research requires the expertise of many kinds of scientists including biostatisticians, molecular biologists, computer specialists, social workers and geneticists collaborating with a network of physicians at New York Presbyterian Medical Center and other clinics and medical centers all over the country.

#### **Findings from our study include:**

1. Identification of BRD2 on chromosome 6, the first gene identified for JME and the first for any IGE.
2. Demonstration that different IGEs have different genetic influences.
3. Maternal inheritance in JME
4. Identification of ME2 on chromosome 18, a gene involved in several different IGEs
5. Demonstration of the genetic basis of JME is different in Caucasians and non-Caucasians

**BACKGROUND:** In 1981, Dr. David Greenberg started the Epilepsy Genetics Project with the Comprehensive Epilepsy Center in Los Angeles, California. The goal of the project was to identify genes involved in an epilepsy syndrome called Juvenile Myoclonic Epilepsy (JME or Janz Syndrome, first identified by Dr. Dieter Janz). JME is non-progressive epilepsy that occurs in families and begins during adolescence. In 1987 this group published the first finding of a gene location for any form of epilepsy. This finding was the first proof of the genetic basis of Idiopathic Generalized Epilepsy. Subsequent studies, including an independent analysis from Dr. Janz’s laboratory, confirmed this finding. In the research community, this study serves as a model to approach diseases with strong genetic influence.

In 1990, the Epilepsy Genetics Project moved to New York City to begin collaboration with 3 area medical institutions continuing at the forefront of genetic research for JME, other IGEs and complex common diseases. By 1996, the Epilepsy Genetics Project, re-named the Family Studies in Epilepsy Program, expanded beyond New York City. Today it includes more than 100 collaborators nationwide and has recruited more than 350 subjects and family members. We explore adolescent and childhood onset IGEs that run in families. They include Juvenile Myoclonic Epilepsy (JME), Juvenile Absence Epilepsy (JAE), Childhood Absence Epilepsy (CAE), Epilepsy with Awakening Grand Mal seizures (AGM), Epilepsy with Random Grand Mal Seizures (RGM), and Benign Rolandic Epilepsy (BRE) childhood onset focal epilepsy thought to be genetic.

**ARE YOU A CANDIDATE?** We are seeking patients diagnosed with the Idiopathic Generalized Epilepsies listed above. African-American and Hispanic families are of particular interest in our research because IGEs in minority groups in the USA have not been well studied. We hope to collect data on as many subjects as possible, as well as their family members. Your physician can provide you with additional information about our studies.

If you think you are a candidate, please call Ms. Irene Klotz, Director of Recruitment, 212-342-0489 or email us at [rene@shallot.cpmc.columbia.edu](mailto:rene@shallot.cpmc.columbia.edu). You may also contact our Study Associates, Nicole Iorillo and Janine Rose at 212-342-0482-3.

Participation will not interfere with normal medical care. Subjects and family members will be contacted only once or twice for 1-2 hours, usually by telephone. We interview each subject to obtain a clinical/family history of epilepsy and ask each participant to provide a small sample of saliva using a special container that we provide. All testing is free of charge.