KCNQ2-related Epilepsy and Neurodevelopmental Disorders Patient Registry
The RIKEE Network

What are KCNQ2-related disorders?

KCNQ2-related developmental disorders range from quite mild to severe. They are most commonly seen when a child has a change (also called a *mutation* or *variant*) in the KCNQ2 gene. Variants in KCNQ3 and KCNQ5 can also cause disease. These genes control the function of potassium channels in the brain. So far, only a small number of patients who have such variants have been diagnosed. Many patients with KCNQ2-related disorders remain undiagnosed. Even after diagnosis, many are not reported in the medical information available to doctors and the public. Not knowing more about the range of KCNQ2-related disorders continues to make it harder to help newly diagnosed patients and their families.

What is the RIKEE registry?

The RIKEE Network is a group of doctors and scientists working to understand and better treat KCNQ2-related disorders, including epilepsy and slow development. A "registry" is an organized system to collect information about a condition. By enrolling your child in the RIKEE registry, you will be contributing to efforts by doctors, scientists, and family-led groups to understand KCNQ2-related illnesses better and develop new treatments. Participation is completely voluntary.

How can I participate?

If your child has been diagnosed with a variant in KCNQ2, KCNQ3, KCNQ4, or KCNQ5, or your family is known to have epilepsy in newborn babies, you can get more information and enroll in the registry by emailing kcnq2info@bcm.edu. You can also speak with your treating neurologist who can enroll you anonymously in the registry if you give them verbal permission.

What can I expect if I participate?

If you choose to participate in the registry, your file will first be assigned a research code number. We will also ask your permission to look in the patient's medical record to gather additional information we may need for the research. All labels directly identifying you (name, address, date of birth, etc.) are removed, but remaining medical information will be added in the registry. Your information will be grouped with other participant information so that researchers and doctors can better understand patterns of KCNQ2-related illness. The results will be discussed in scientific articles and conferences, and used for planning new research and treatments.

Www.RIKEE.org is a website making medical information on KCNQ2/3/5 variants available to families, researchers, doctors and others counseling patients. You will be given the option to allow your child's or your family's limited information to be included in this website's list of variants. There will be no information included that will identify you.

Questions? Email us at kcnq2info@bcm.edu Website: www.RIKEE.org