What are neonatal seizures?
Seizures are attacks involving loss of awareness and body control that come on suddenly and involuntarily. Seizures can come on at any age, including in the days after birth. For most babies with seizures, a cause can be determined by testing performed in the hospital, such as blood tests and brain magnetic imaging scans. The required testing differs from patient to patient and is chosen by the hospital team. Treatment depends on the cause found.

What are genetic causes of neonatal-onset epilepsy?
Doctors diagnose “epilepsy” when seizures continue in a patient. In some babies with seizures, routine blood and brain scan testing in the hospital shows no cause for seizures, but DNA testing performed in specialized clinical genetics labs can reveal changes in a gene that are the cause of the epilepsy. Some of the changes known to cause epilepsy in babies are found in the genes KCNQ2, KCNQ3, and SCN2A. Changes in other genes can also do this.

What is ERGENT?
A group of doctors have been caring for patients with genetic forms of epilepsy and studying the genes to understand why they cause seizures and other effects on young children. The research team made lists of the features that babies with neonatal onset genetic epilepsy seem to have in common. ERGENT is a study to test whether these features can be used to predict which babies have genetic epilepsy, soon after birth and even before the testing is performed.

What can I expect if I participate?
If you choose to participate in ERGENT, your doctor will arrange for a no-cost blood test of over 180 genes known to cause epilepsy. The test results will be sent to your doctor (and to you, if you request that). We will also ask your permission to review the patient’s medical records (including the genetic testing) to gather information about their medical condition and treatment. All information will then be analyzed. The results will be discussed in scientific articles and conferences, and used for planning new research, diagnostic approaches, and treatments. We have also created a website, www.ergent.org, including information about the study.

WHAT is RIKEE?
RIKEE is a collaboration of doctors and scientists that has been working to understand and develop better treatments for babies with genetic causes of epilepsy. Our research plan, including ERGENT, has been reviewed and approved by the Institutional Review Board at Baylor College of Medicine in Houston, TX. The lead investigator is Edward Cooper, MD PhD.

ERGENT has been funded by three non-profit organizations led by parents of children with neonatal onset genetic epilepsies: the Jack Pribaz Foundation, the KCNQ2 Cure Alliance, and the FamilieSCN2A Foundation.