Questions and Answers with Dr. Darryl De Vivo and Kris Engelstad

Can you tell us more about the lab environment at Columbia?

The Colleen Giblin Laboratory currently has six staff members, in addition to Dr. De Vivo, who work on Glut1 DS related projects in basic research. The Staff includes three MD's, one PhD, and two people with MS degrees. We collaborate with investigators in the Motor Neuron Center, Columbia Pathologists, and Columbia Radiology, as we develop outcome measures in Glut1 DS mice. We conduct studies in cultured cells, and in mutant mice that we created in 2006. The mouse model allows us to pursue symptomatic and disease-modifying treatments for Glut1 DS.

How is your research funded?

We are funded by the National Institutes of Health, several pharmaceutical industries and private foundations.

What is your lab working on at this time?

We are working on a variety of projects to characterize the neurological deficits in Glut1 deficient mice and to evaluate various treatment options that may eventually be useful in humans.

When will the proposed Glut1 Deficiency studies begin (or when did they begin)?

Glut1 DS studies began in 1991 when we described this disease in two infants, and continues to the present time as we search for better treatments.

When do you expect them to be completed and the results shared?

We expect our studies to be completed when we develop a cure for this disabling condition, and not a minute sooner.

What potential does this research have to impact the big picture for Glut1 Deficiency (and perhaps even more conditions)?

Our current research specifically addresses the ongoing needs of patients and their families who struggle with the daily challenges associated with Glut1 DS. But, we always hope that focused research on one rare disease will be relevant to other diseases in the long run, and we have many examples where this hope, in fact, has been realized.

How does this project reflect your overall approach to research and medicine?

Child Neurologists have cared for children with rare diseases from the beginning, witness Bernard Sachs and Tay-Sachs disease. Professor Sachs was the first Director of Child Neurology at Columbia University (1934). There are now about 7000 rare diseases known today, and 80% are genetically determined. Two-thirds of these rare diseases affect children, and 30% of these children succumb to their disease by age 5 years. Glut1 DS is an excellent example of a rare disease, described at Columbia University in 1991, also genetically determined, and affecting infants children, and adults. Our approach to research and medicine always has been translational (even before the term became fashionable). The process always starts with the patient, moves to the laboratory, and later returns to the patient as we search for better treatments and cures. Our laboratory team conducts research on patient blood samples, cultured cells, and animal models of the disease. Our clinical team conducts IRB approved research on patients while simultaneously providing state-of-the-art clinical care. The team is multi-disciplinary and includes child neurologists, epileptologists, movement disorder specialists, geneticists, physical therapists, genetic counselors, nutritionists, and clinical coordinators. These team members devote their lives to the care of patients with rare diseases including Glut1 DS, and to the conduct of clinical and basic research that will inform us of the disease mechanisms and the pathophysiology. We believe that this model represents translational medicine at its best, and it is faithful to the basic principle that "today's research is tomorrow's treatment ".

What can the Glut1 Deficiency community do to be helpful to you and where can we find more information?

Our translational approach to the care of Glut1 DS patients and other children with rare diseases is an excellent medical model, but it is a bad business model. To sustain our multidisciplinary approach to patient care and to conduct important translational research, supplemental funding is imperative. We continue to seek supplemental funding from federal and non-federal agencies and from philanthropic contributions. The more time we spend seeking support, the less time we have to provide patient care and to conduct clinical research. Financial security of the translational enterprise increases the likelihood of improving patient care and finding the elusive cure. Funding, in essence, is the fuel that determines the speed with which we reach our shared goal. And, the patients are the ultimate beneficiaries of your generosity.