Investigator Spotlight: Jeffrey Neul, MD PhD, Jan and Dan

Duncan Neurological Research Institute at Texas Children's Hospital and Baylor College of Medicine

With winter having passed by quickly and spring in the air, IRSF is busily preparing for the World Rett Syndrome Congress taking place this June. With that in mind, we are eager to highlight the dedicated scientists who have planned this highly anticipated event to be held for the first time in the US in New Orleans, LA. This month we are pleased to move the spotlight onto Dr. Jeffrey Neul from the Jan and Dan Duncan Neurological Research Institute at Texas Children's Hospital and Baylor College of Medicine in Houston, TX. Dr. Neul is the Chair of the Translational and Pre-Clinical Research Symposium at the World Congress. He has put together an exciting program where current knowledge in translational and clinical research on Rett syndrome will be presented and discussion on methods to accelerate these fields will be encouraged.



Dr. Jeffrey Neul is a physician scientist and had received his MD and PhD in Developmental Biology from the University of Chicago. He joined Baylor College of Medicine for a Pediatric Internship and a Residency in Child Neurology. Dr. Neul is now an Associate Professor in the Departments of Pediatrics – Section of Neurology at Baylor College of Medicine. He also holds positions in the Department of Molecular and Human Genetics, Molecular Physiology and Biophysics, and Neuroscience. Dr. Neul is the Anthony and Cynthia Petrello Endowed Scholar of the Jan and Dan Duncan Neurological Research Institute at Texas Children's Hospital. At the Blue Bird Circle Rett Center, he is the Assistant Medical Director and sees patients with Rett syndrome and conducts clinical research on RTT.

In addition to his clinical work, Dr. Neul and his lab are interested in using animal models and molecular techniques to understand the mechanisms that cause some of the specific clinical features found in Rett Syndrome, specifically, in understanding autonomic dysfunction and early death. They have determined that a mouse model reproduces many of the autonomic abnormalities observed in people with Rett syndrome, including breathing problems and abnormal heart rhythm. Dr. Neul and his lab have recently been awarded a HeART grant for "Pharmacological treatment of cardiac rhythm abnormalities in Rett syndrome". Dr. Neul also participates in the Natural History Study for Rett syndrome. In conjunction with this study, IRSF will support him to create a DNA repository for Rett syndrome with the goal in mind to identify genetic mutations other than MECP2 that may contribute to the clinical severity of Rett syndrome.

Besides his work in the clinic and the lab, Dr. Neul was a major participant in IRSF's Rett Syndrome Public Service Campaign featuring Clint Black last year. Given his significant contributions towards IRSF's mission to advance research for treatments and a cure for Rett syndrome while improving the overall quality of life for those living with Rett syndrome today, Dr. Neul was awarded the Circle of Angels Award for Outstanding Research in Rett syndrome at the 2011 Rett Syndrome Education and Awareness Conference.

What prompted you to begin a career in research?

As an undergraduate in chemistry, I became fascinated by the way a small ion such as lithium could have a dramatic effect on behavior and mood, which was the beginning of my desire to do neuroscience and be trained as a physician and a scientist. I became specifically interested in Rett syndrome when during my final year of medical school I asked my child neurology mentor, Peter Huttenlocher, where I should train for child

neurology. He turned, picked up an open journal lying on his desk, and said "Huda Zoghbi identified the gene for Rett syndrome, so you should go work with her." It was one of the best pieces of advice any one has every given me.

What is the single most rewarding aspect of conducting Rett syndrome research?

I love working with the children affected with Rett syndrome and thinking about how we can do research to help them live fulfilling lives.

If you could pick any one symptom of Rett syndrome to prevent or to provide relief for, what would it be?

If I could, I would try to figure out how to let these children talk. I know they have a lot of things they want to tell their families, and being able to communicate would make a remarkable change in their lives.

Besides your role as principal investigator on this project and as a Rett syndrome investigator, what other roles do you currently hold that are specific to the field of Rett syndrome research?

I am the assistant medical director of the Blue Bird Circle Rett Clinic at Texas Children's Hospital and an investigator on the Rett Syndrome Natural History Study. I am on the IRSF Scientific Review Board and the IRSF Medical Advisory Board, and serve on the FDA Orphan Products Division Grant Review Panel.

Provide any other interesting information about yourself or your work that you would like the Rett syndrome community to know about you.

Outside of the lab and clinic, I spend as much time as possible with my wife and two children. I love bicycling and have participated in the MS150 Houston to Austin ride multiple times.

For more information on Dr. Neul, please visit: www.bcm.edu/pediatrics/neurology/?pmid=14799 www.bcm.edu/genetics/index.cfm?pmid=13556