Investigator Spotlight – Dr. Jenny Downs, Telethon Kids Institute in Australia

Dr. Jenny Downs was funded a Neuro-Habilitation HeART award for her project titled “An evaluation of environmental enrichment for young girls with Rett syndrome”. She recently gave us a quick update on the project. Her research aim was to recruit girls with Rett syndrome younger than six years of age and assess the effects of a six-month intensive environmental enrichment program on their gross motor skills, serum levels of BDNF and other aspects of their wellbeing including growth, sleep and mood. Using the infrastructure of the InterRett database, they recruited 12 families from China whose daughters with Rett syndrome participated in the treatment at the Rett Syndrome Comprehensive Research Institute in Shenzhen, China. The girls were on average 3 years of age, each had a pathogenic MECP2 mutation and mutations associated with milder and more severe clinical severity were represented. The collaboration between members of the research team in Perth and China was extremely strong. The program was well received by the families and appeared to be enjoyed by the participating girls. The last two weeks of the program were dedicated to ensuring that the parents had the skills and understanding for how they could encourage continuation of their daughter’s activities after the program ended. Dr. Downs and her colleagues are currently writing a manuscript of their findings, and thereafter look forward to opportunities for their dissemination.

Dr. Steve Kaminsky comments, “This is a unique study that will demonstrate whether active physical therapy in an enriched environment will enhance the motor sensory of children with Rett syndrome. We look forward to sharing the study’s results when they are published. ”

We asked Dr. Downs to complete a questionnaire at the start of the study so our community could get to know her.

1) What prompted you to begin a career in research?

I have always felt passionate about wanting to be able to better understand neurodevelopmental disorders, and importantly, to better understand how best to manage any problems in order to achieve the best outcomes. I previously managed to combine research projects with my clinical physiotherapy work with children when seeking answers to clinical issues. But research takes a lot of time, and it was not until the opportunity arose to work with Dr. Helen Leonard that I was able to become more deeply involved in exciting research projects to help Rett syndrome.

2) Provide a brief outline of your training and the work you have conducted that has led to this proposal.

I have a strong interest in the benefits of movement and exercise for both children and adults and particularly for those with a neurodevelopmental disorder. My background is in both research and clinical practice and I have been able to work on projects in Rett syndrome, many of which relate to early development, functional abilities, physical activity and scoliosis. There is now increasing evidence that learning and development is favored in an enriched environment containing lots of physical activity, and some of this evidence relates specifically to Rett syndrome. I am very excited to be able to run this study which will investigate the effects of an enriched environment program for young girls in China who are newly diagnosed with Rett syndrome.

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

For me, the single most rewarding aspect of conducting Rett syndrome research is linking research findings with the practical needs of these amazing girls, women and their families.

4) Identify a potential positive outcome of the research you are conducting that is specific to this proposal. (i.e. Does this project target a specific symptom of Rett syndrome?)

This study could find that gross motor skills are improved following participation in an intensive environmental enrichment program.

5) What other disease(s) does your research focus on?

Most of my research relates to Rett syndrome but I also have projects underway on preterm infant development, the CDKL5 disorder and MECP2 Duplication Syndrome, early onset scoliosis and Duchenne muscular dystrophy.