This was a global meeting of leaders in the areas of Rett syndrome research, clinical care, and the role of parent associations. It was a scientific symposium plus family support, education and awareness conference all rolled into one. The meeting combined plenary talks with multiple concurrent scientific sessions and workshops, complemented by research poster boards and parent association information booths.

Over the course of 4 days, many presentations were delivered demonstrating where research has brought us in the nine years since discovering the genetic cause of Rett syndrome. Many of the sessions were recorded, and after final permissions are secured, these sessions or handouts will be available online for everyone to view.

The meeting opened with talks given by Drs. Huda Zoghbi and Adrian Bird, two researchers who have made seminal discoveries in the field. Dr. Zoghbi along with other investigators in 1999 identified the MECP2 gene as responsible for the majority of cases of Rett syndrome. Since that time her work has been devoted to understanding the function of MeCP2 with a focus on interactions with other nuclear signaling molecules. Dr. Adrian Bird gave an overview of the structure and function of MeCP2 and described how MeCP2 acts as a "brake" on gene expression. He focused on showing precisely how MeCP2 interfaces with DNA in order to repress the transcription of genes. Dr. Bird also discussed his lab's finding in 2007 that Rett Syndrome is potentially reversible through the work done using specific mouse models of the disease.

Using genetic Mecp2 "knockout" mouse models has uncovered the underlying neurological changes of those affected by Rett syndrome. This meeting also shows the important need for new mouse models that more precisely model the different mutations that occur in girls with RTT. The conference highlighted how much more is now known about the role and function of the MeCP2 protein in human nervous system development. It follows then that if we can fully understand the role of MeCP2 when it is working correctly, we will have a better path towards repairing, bypassing, or overriding dysfunctional forms of MeCP2. Part of the meeting focused on relationships between genotype and phenotype - essentially determining how different mutations and modifiers appear to impact the severity of RTT. The meeting also outlined the range of Rett syndrome severity; females and males show differences in age of onset, degrees of severity and there is a wide spectrum of different presentations, from mild learning disabilities to classic RTT. Mutations that arise in genes other than MECP2- such as CDKL5 and FOXG1 - are also now implicated in causing a Rett-like disorder. These might shed light on pathways that are upstream or downstream of MeCP2.

All of this information increases our understanding of the disorder and leads researchers in new directions. Several presentations and sessions were offered around medical issues and clinical/therapeutic care in Rett syndrome, including seizure/epilepsy management; breathing issues; scoliosis and bone health; and gastro-intestinal problems. The
benefits of therapies ranging from physical therapy to music therapy and communication strategies were also addressed.

The Internet is emerging as a valuable tool for researchers to gather data on rare diseases. Our own Dr. Helen Leonard discussed the AussieRett and InterRett online databases, which have greatly expanded clinical research on Rett syndrome. Dr. Leonard's presentation discussed the great value in combining data from different sources through initiatives such as the international database InterRett. Through the use of this internet database, families and clinicians from around the world can contribute their information and help researchers gather enough data to make statistically relevant analyses possible.

An overriding take-home message from this Congress was the need for ongoing international collaborations. Time and funds are not put to their best when we work in isolation, or too competitively. No single lab or clinic sees enough patients to reach statistically meaningful numbers. In order to launch real clinical trials, we need to build shared datasets, make clinical assessments using the same criteria, and keep the needs of those affected by Rett syndrome and their families at the centre of the work. In the concluding talk, Dr. Carolyn Schanen summarized some potential treatments that are on the horizon for those with Rett syndrome. These include new compounds that seek to target non-sense mutations, gene-based therapies and other therapies that target neuronal growth factor systems.