Current Research & Future Possibilities

Dr. James Eubanks

At the Building Bridges Conference held in Kingston, Dr. Eubanks presented several research highlights stemming from presentations made at international conferences by a number of scientists investigating different aspects of Rett syndrome. The conferences included the 2nd European Working Group on Rett Syndrome held in Stresa Italy in October 2009, a Strategic Planning Workshop sponsored by the International Rett Syndrome Foundation held in New York City in March 2010, and the 11th Annual Rett Syndrome Research Symposium held in Washington DC in June 2010.

The topics of the meeting in Stresa included studies from prominent Rett syndrome investigators such as Adrian Bird, Yi Sun, David Katz, Gail Mandel, John Bissonnette, Monica Justice, John Christodoulou, Tommaso Pizzorusso, and Peter Huppke amongst others. Dr. Eubanks reviewed the results that were presented, and explained how they affected the search for treatments for Rett syndrome. Specific highlights include: (1) the demonstration that raising MeCP2-deficient mice in an enriched environment improves their phenotype and strengthens the ability of their neurons to communicate with one another; (2) that genes other than MeCP2 contribute to the severity of Rett syndrome, and that some of these modifier genes are beginning to be identified; (3) That neural circuits in MeCP2-deficient mice are hypersensitive, and that certain drugs are able to normalize the hypersensitivity; and (4) that at least some of the breathing deficiencies present in MeCP2-deficient mice can be corrected with specific drugs.

The topics of the meeting in New York City included the interests of IRSF to foster partnerships with the National Institutes of Health to facilitate drug development and implementation for Rett syndrome - which are moving forward and now appear likely to happen; (2) the announcement of new research competitions to be funded by IRSF that are dedicated to developing treatment strategies for Rett syndrome; and (3) developing a means to bring medicinal chemists who understand how to make specific drugs together with basic scientists who know what types of drugs need to be developed to treat Rett syndrome. The focus of the meeting was to jump-start pathways to getting drugs tested first in mice, and then if the results are encouraging, developing means to get these drugs into clinical trials. This meeting was the necessary first step in the process.

The topics of the meeting in Washington DC were broad in nature, but the primary take-home message was that Rett syndrome is a treatable condition - we now need to determine which strategies and which drugs are the best routes for use in the clinic. A number of investigators presented results in MeCP2-deficient mice showing that their Rett-like condition could be improved by genetic procedures related to gene therapy, with drugs that target the BDNF pathways in the body, and by drugs that act to calm certain hyperactive sites in the brain. Each of these studies was viewed as encouraging by those in attendance, and although none of the procedures are presently approved for clinical use, the overriding consensus was that the field is moving forward towards developing treatments, and that more drugs will soon be ready for testing in clinical trials.