# Press Release:
# Rett Syndrome Research Trust Creates MECP2 Consortium

***September 7, 2011***

**Media Contact:**
*Monica Coenraads*
*Executive Director, RSRT*
*monica@rsrt.org*
*203.445.0041*

READ ACCOMPANYING INTERVIEW ON THE RSRT BLOG

*SPANISH TRANSLATION*
*GERMAN TRANSLATION*
*ITALIAN TRANSLATION*

# Rett Syndrome Research Trust Creates MECP2 Consortium

The Rett Syndrome Research Trust (RSRT) announced today the creation of the MECP2 Consortium, a dynamic collaboration between the laboratories of three distinguished scientists, Adrian Bird, Michael Greenberg and Gail Mandel. Made possible by a $1 million gift by RSRT Trustee Tony Schoener and his wife Kathy, the Consortium was formed to definitively determine how this complex protein, MeCP2, functions and exerts its powerful influence on the human brain. Deep knowledge of its structure and roles in neurological development and maintenance will inform the design of rational treatments for disorders caused by abnormalities in this protein. RSRT will invest $1.8 million in the Consortium.

Alterations in the methyl CpG-binding protein (MeCP2) cause the autism spectrum disorder Rett Syndrome and the recently identified MECP2 Duplication Syndrome, and are seen in some cases of classic autism, childhood schizophrenia and milder neuropsychiatric conditions such as anxiety and learning disabilities.

Rett Syndrome strikes little girls almost exclusively, with first symptoms usually appearing before the age of 18 months. These children lose speech, motor control and functional hand use, and many suffer from seizures, orthopedic and severe digestive problems, breathing and other autonomic impairments. Most live into adulthood, and require total, round-the-clock care. The core symptoms of the MECP2 Duplication Syndrome include developmental delay, absent to minimal speech, recurrent infections, progressive spasticity, ataxia, autistic features, and seizures.

The Consortium members have each made novel contributions to the MeCP2 field. Adrian Bird of the University of Edinburgh discovered the MeCP2 protein in the early 1990's, and authored the seminal 2007 Science paper which introduced the startling possibility that Rett Syndrome and other MeCP2 related disorders are curable. Michael Greenberg, Chairman of the Department of Neurobiology at Harvard University, identified and demonstrated the importance of phosphorylation of MeCP2 at specific sites in response to neuronal activity. Gail Mandel, a Howard Hughes Investigator at Orgeon Health and Sciences University, discovered an unexpected and central role for glial cells in the etiology of Rett Syndrome.

Despite ongoing efforts by dozens of labs spanning two decades, the function of the MeCP2 protein remains elusive. "The Consortium is RSRT's response to solving a complex issue: convene thought leaders who possess distinct but synergistic expertise; facilitate the necessary infrastructure to enable real-time sharing of information; provide the necessary financial support to give each investigator the freedom to follow their scientific instinct. We are indebted to the Schoeners for making the Consortium possible," said Monica Coenraads, Executive Director of RSRT and parent of a teenaged daughter severely disabled by Rett Syndrome.

Professors Bird, Greenberg and Mandel jointly comment, "This is a new and exciting experience for all of us. Our laboratories, like many in academia, have mostly worked solo. Now we are 'opening our books' to the Consortium, sharing information about Rett Syndrome research and collectively generating new ideas. By putting our heads together we want to accelerate research on a key question that has proven difficult to answer up to now: what exactly does MeCP2 do for the brain? The answer will have profound implications for attempts to treat disorders caused by mutations in the MECP2 gene. Now feels like the right time to push forward on this - the Consortium gives us a golden opportunity to make real progress."

"Hearing Dr. Greenberg state at the Consortium's recent meeting, 'This conversation just saved me a year's worth of work' highlights the crucial need for financially and intellectually facilitating such close collaborations. We are delighted that our funds have helped launch this effort and are optimistic that our investment will be leveraged many times over," said Tony and Kathy Schoener, parents of an 18-year-old daughter with Rett Syndrome.

To read an interview with the Consortium members please **visit RSRT Blog.**

# Rett Syndrome Research Trust Creates MECP2 Consortium

### *About Rett Syndrome*

Rett Syndrome is a genetic neurological disorder that almost exclusively affects girls. It strikes randomly, typically at the age of 12 to 18 months, and is caused by random mutations of the MECP2 gene on the X chromosome. Rett Syndrome is devastating as it deprives young girls of speech, hand use, normal movement often including the ability to walk. As the girls enter childhood the disorder brings anxiety, seizures, tremors, breathing difficulties, severe GI issues. While their bodies suffer, it is believed that their cognitive abilities remain largely intact. Although most children survive to adulthood, they require total round-the-clock care.

### *About the Rett Syndrome Research Trust*

RSRT is a non-profit organization with a highly focused and urgent mission: eradicate Rett Syndrome and related MECP2 disorders. In search of a cure and effective treatment options, RSRT operates at the center of global scientific activity, funding bold projects that are unlikely to be supported by the NIH or other more traditional funding agencies. RSRT refutes the conventional practice of labs working in isolation, instead seeking out, promoting and funding collaborations and consortia in which scientists work across multiple disciplines. These relationships enable the development and execution of a research agenda that neither academia nor industry could achieve alone. Since 2008, RSRT has provided $25 million of financial support to: 4 clinical trials testing 3 compounds, 33 scientists in 27 academic institutions and 3 biotech firms. To learn more about the Trust, please visit **www.ReverseRett.org**.