

Press Release:
The Rett Syndrome Research Trust
Launches Operations

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The Rett Syndrome Research Trust, a newly established nonprofit organization, is pleased to announce the launch of our operations and website (www.rsrt.org). In the wake of last year's dramatic pre-clinical reversal of Rett symptoms, the Trust has been formed to pursue the next steps toward treatment and cures for Rett Syndrome and the increasing range of MECP2 spectrum disorders. Our short-term goal is to bring novel therapeutics addressing the underlying MECP2 pathology to clinical trials within five years.

Rett Syndrome is the most physically disabling of the autism spectrum disorders and the leading cause of severe genetic impairment in females, with an estimated 30,000 individuals afflicted. At this time the number of children born with other MECP2 spectrum disorders remains unknown. In the U.S., the frequency of autism has risen to 1 in 150 children.

Classic Rett Syndrome affects females almost exclusively. Symptoms typically begin to appear in little girls between infancy and two years of age. Onset may be heralded by hypotonia and delayed development, or by regression and loss of acquired skills, including speech and functional hand use. As the disorder progresses, symptoms may include disordered breathing, sleep disturbances, severe digestive problems, orthopedic abnormalities, anxiety, apraxia, seizures, impaired cardiac and circulatory function and Parkinsonian tremor. Many children with Rett Syndrome are wheelchair bound. Most victims survive well into adulthood; all require total, 24-hour-a-day care. The cost, both financial and in terms of human suffering, is enormous.

The vision of the Rett Syndrome Research Trust is guided by founders and advisors who have been responsible for many of the major advances in Rett research over the past decade. Based on their comprehensive analysis of strategic priorities at our inaugural international meeting, RSRT's initial projects build the foundation for a drug discovery platform. Our first five projects represent diverse yet complimentary approaches to treatment and reversal, encompassing drug screening, elucidation of target areas, and control of MECP2.

The projects include an international collaboration sponsored jointly with Autism Speaks and the Medical Research Council (MRC) of the UK. The collaboration is based in the laboratory of Adrian Bird, Ph.D., at the University of Edinburgh, with key contributions from Nathaniel Heintz, Ph.D. of Rockefeller University. Professor Bird, who first isolated the MeCP2 protein in 1992, last year published his astonishing reversal of full-blown symptoms in a model of Rett Syndrome. He is the Buchanan Professor of Genetics at Edinburgh, and Deputy Chairman of the Wellcome Trust. Dr. Heintz is the James and Marilyn Simons Professor of Molecular Biology, and a Howard Hughes Medical Institute Investigator whose work involves identification of novel pathways that participate in the development, function and dysfunction of the mammalian brain. Dr. Heintz has pioneered a variety of advanced genetic tools that can be employed to understand the impact of Rett Syndrome on specific brain circuits. This collaboration will inform all potential translational research, whether through gene therapy, protein replacement or drug discovery.

"We are extremely pleased to be collaborating on this important effort. The exciting recent discoveries by Rett Syndrome researchers have energized the field. This collaboration will help sustain its momentum," says Geri Dawson, Chief Science Officer at Autism Speaks.

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Huda Zoghbi, M.D. whose 1999 research breakthrough revealed that Rett Syndrome is caused by mutations in the MECP2 gene, is a key advisor to RSRT and Professor in the departments of Molecular and Human Genetics, Pediatrics, Neurology and Neuroscience at Baylor College of Medicine and a Howard Hughes Medical Institute Investigator. She is particularly interested in the growing number of clinical presentations of MECP2 spectrum disorders, which include autistic profiles, and their molecular underpinnings. She comments on the relevance of this work to conditions beyond classic Rett Syndrome: “Rett Syndrome and MECP2 disorders provide an opportunity to understand the neuronal and molecular basis of a variety of neuropsychiatric disorders. Patients with Rett and related disorders manifest features of so many of the neurological disorders we encounter in the clinic. Thus, understanding the pathogenic mechanism underlying MECP2 disorders is likely to help us understand diseases ranging from autism and schizophrenia to cognitive disorders, epilepsy, and Parkinson disease.”

Monica Coenraads, Executive Director of the Trust, describes the work ahead. “This early stage of drug discovery is unlikely to be a high funding priority for the National Institutes of Health, large pharmaceutical or small biotech companies, but is the Trust’s sole and immediate pursuit. RSRT has been structured as an agile, science-driven organization, able to respond dynamically to emerging research. A private effort is essential if treatments and cures are to be developed in a time frame that the devastating nature of this disorder demands.”

For more information on Rett Syndrome, MECP2 spectrum disorders and the first projects the Trust has undertaken, please visit the website of the Rett Syndrome Research Trust at rsrt.org.

About Autism Speaks

Autism Speaks is dedicated to increasing awareness of autism spectrum disorders, to funding research into the causes, prevention and treatments for autism, and to advocating for the needs of individuals with autism and their families. It was founded in February 2005 by Suzanne and Bob Wright, the grandparents of a child with autism. Bob Wright is Senior Advisor at Lee Equity Partners and served as vice chairman, General Electric, and chief executive officer of NBC and NBC Universal for more than twenty years. Autism Speaks merged with both the National Alliance for Autism Research (NAAR) and Cure Autism Now (CAN), bringing together the nation’s three leading autism advocacy organizations. To learn more about Autism Speaks, please visit autismspeaks.org.

About the Medical Research Council

The Medical Research Council (MRC) is a publicly funded UK organization dedicated to improving human health. It supports research across the entire spectrum of medical sciences, in universities and hospitals, in their own units and institutes in the UK, and in Africa. In the past year MRC supported nearly a billion dollars of research. To learn more about the MRC, please visit www.mrc.ac.uk.