

Press Release:
Rett Syndrome Research Trust Funds
Innovative Research at UCSF to Accelerate
Drug Discovery

August 24, 2009

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

Rett Syndrome Research Trust Funds Innovative Research at UCSF to Accelerate Drug Discovery



The Rett Syndrome Research Trust announced today its financial support of an innovative research project with significant therapeutic potential. The project will be conducted in the laboratory of Stavros Lomvardas at the University of California, San Francisco.

Lomvardas discovered that the MeCP2 protein, associated with a variety of neurological disorders, including Rett Syndrome, regulates olfactory neuron differentiation in an unusual way. Olfactory receptor neurons, or ORNs, are crucial to our sense of smell. A differentiated, mature olfactory neuron expresses several molecular markers in a mutually exclusive manner.

But Lomvardas observed that in MeCP2 knockout mice these neurons co-express these molecular markers. Lomvardas will capitalize on the violation of the mutually exclusive expression pattern to develop a molecular assay for MeCP2 activity. The assay will then be used in a high throughput screen to identify chemical compounds that can reverse the molecular consequences of MeCP2 deletion in olfactory neurons. These experiments may be consequential to disease areas beyond Rett where epigenetic phenomena play a role.

“To my knowledge there are currently no cellular assays for MeCP2 dysfunction that are amenable to high throughput screening. Dr. Lomvardas is therefore uniquely positioned to develop the first in vitro assay that can be utilized to screen tens of thousands of chemical compounds in search of novel drugs to treat Rett Syndrome. Furthermore his research has the potential to contribute significantly to our understanding of MeCP2 function,” said Adrian Bird, RSRT trustee and scientific advisor.

Lomvardas received his doctorate degree in biochemistry at Columbia University where he trained in the laboratory of Dimitris Thanos and Nobel Laureate Richard Axel. He established his independent lab at UCSF two years ago. Though at an early stage in his career, Lomvardas has already generated an impressive list of high-profile manuscripts.

“It is a great honor to be selected for funding from the Rett Syndrome Research Trust. I am fully committed to the goals of the Trust and I will work hard towards the better understanding of the causes of Rett Syndrome and the development of potential therapeutic approaches,” said Lomvardas.

“Dr. Lomvardas is precisely the kind of out-of-the-box thinker and productive scientist RSRT seeks to recruit into the field. He has a track record of generating and executing on transformative ideas. I look forward with anticipation to the advances he will undoubtedly make,” said Monica Coenraads, executive director of the Rett Syndrome Research Trust and mother to a daughter with the disorder.

An interview with Dr. Lomvardas is available online at the RSRT Blog.

Rett Syndrome Research Trust Funds Innovative Research at UCSF to Accelerate Drug Discovery



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.

Our Partners

Our partners in supporting this work are parents' organizations worldwide including [Reverse Rett \(UK\)](#), [Rett Syndrome Research & Treatment Foundation \(Israel\)](#), [Skye Wellesley Foundation \(UK\)](#), [Rett Syndrome & CDKL5 Ireland](#), [Rett Syndrom Deutschland](#), [Stichting Rett Syndrome \(Holland\)](#).

Our U.S. partners that helped make this research possible include [Girl Power 2 Cure](#), [Eva Fini Fund at RSRT](#), [Kate Foundation for Rett Syndrome Research](#), [Rocky Mountain Rett Association](#), [Anastasi Fund](#), [Claire's Crusade](#), [New Jersey Rett Syndrome Association](#), [Rett Syndrome Association of Massachusetts](#), and the [MECP2 Duplication Syndrome Fund](#) at RSRT.