

## IRSF Awards \$1.5M for Research

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The International Rett Syndrome Foundation Awards \$1.5M for Novel Basic and Translational Research Programs

\$30M cumulative research dollars have been awarded in a quest to accelerate treatments

Cincinnati, (OH) - The International Rett Syndrome Foundation (IRSF) broadened and intensified its efforts to produce treatments for Rett syndrome and related disorders, announcing today that it is awarding another \$1.5M to support 18 new grants at leading global research institutions. In 2012, IRSF invested nearly \$4M in high-quality, peer-reviewed research grants that give promise to advancing treatment strategies of Rett syndrome and other MECP2 disorders. The grants awarded today are designed to study a variety of diverse topics from basic discovery science and disease pathology to testing compounds and developing outcome measures for Rett syndrome. IRSF is the world's largest private source of funding for biomedical and clinical Rett syndrome research. Since 1998, IRSF has funded \$30M in innovative research grants which have produced critical breakthroughs to advance the understanding of this neurological condition.

IRSF funds pioneering studies in Basic Research that aim to discover new avenues that may be amenable to novel therapies. IRSF also seeks to bridge the critical translational funding gap by investing in Translational Research that includes both pre-clinical and clinical studies. In an effort to recruit future Rett researchers, IRSF creates fellowships to outstanding post-doctoral research scientists and clinical scientists.

The awarded basic research projects cover discovery topics from epigenetics and gene regulation to understanding pathways and other neuronal cell types in Rett syndrome. Together, these studies will allow for identification of new therapeutic targets in Rett syndrome. The second round of translational grants is focused on gene therapy approaches, development and testing of potential therapeutic compounds, and development of outcome measures in humans that will be used in future clinical studies.

"This year's call for proposals produced our largest number of outstanding quality grant applications to IRSF in the history of the organization," commented Steven Kaminsky, PhD, the Chief Science Officer of IRSF. "The astounding response from the science community to our call bodes well for Rett syndrome research and illustrates the growing number of researchers exploring the biology surrounding Rett syndrome and possible treatments that will modify its course." Dr. Kaminsky added, "As more and more fundamental discoveries are made and translated into pre-clinical and clinical trials each year, we get closer to our goal of modifying the biology surrounding Rett syndrome and improving the lives of our Rett patients and their families. With the help of a superb group of reviewers, we have built a plan that funds the best of the best. We look forward to working with these investigators as we continue to push our mission to reverse Rett syndrome."

New Translational Research Awards

- Kevin Foust, PhD, Ohio State University

AAV Gene Therapy for MECP2 Duplication

- Steven Gray, PhD, University of North Carolina at Chapel Hill

MECP2 gene transfer using novel RTT-specific rAAV vectors

- Daniel Glaze, MD, Baylor College of Medicine

Autonomic Nervous System (ANS) Dysregulation in Rett Syndrome: Objective Measures through Pupillometry and ANS Questionnaire

- Kevin Jones, PhD, University of Colorado-Boulder

A Screen for Compounds that Regulate BDNF Expression

- Jay Shapiro, MD, Kennedy Krieger Institute

Treatment of Osteoporosis in Murine Rett Syndrome Models: A Comparison of Zoledronic Acid vs. Teriparatide on Osteoblast Function, Gene Expression and Bone Mass

- Dag Yasui, PhD, University Of California-Davis

Investigation of CHRNA7 Ligands as Potential Rett Therapies

#### New Basic Research Awards-Regular Research Grants

- Chinfei Chen, MD, PhD, Children's Hospital Boston

Testing for Reversibility of Sensory System Circuitopathy in Mouse Models for RTT

- Yvonne Nsokika Fondufe-Mittendorf, PhD, University of Kentucky-Lexington

The Epigenetic Control of Gene Expression by MeCP2

- Ali Khoshnan, PhD, California Institute of Technology

The Role of IKKb Signaling Pathway in Rett Syndrome

- Charlotte Kilstrup-Nielsen, PhD, University of Insubria

Investigation of the Importance of a Hitherto Uncharacterized MeCP2 Phospho-isoform for Neuronal Morphogenesis and Chromatin Related Functions

- David Lieberman, MD, PhD, University of California, San Diego

A Proteomics Based Approach to Restore Bidirectional Homeostatic Plasticity in MeCP2 Deficient Neurons In Vitro and In Vivo

- Alysson Muotri, PhD, University of California, San Diego

Contribution of Human Astrocytes to Rett Syndrome

- Michelle Olsen, PhD, University of Alabama at Birmingham

Altered K<sup>+</sup> Ion and Glutamate Homeostasis in Rett Syndrome

- Beth Stevens, PhD, Children's Hospital Boston

Role of Microglia at Synapses in Rett Syndrome

#### New Mentored Training Fellowships

- Miao He, PhD, Cold Spring Harbor Laboratory

A New Mouse Model for Conditional MeCP2 Inactivation and Reactivation

- Roberto Herai, PhD, University of California, San Diego

A Comprehensive Analyses of Transcriptomic and Proteomic Expression in Rett Syndrome Neurons

- Yun Li, PhD, Whitehead Institute For Biomedical Research

Modeling Rett Syndrome Using TALEN Technology in Human Pluripotent Stem Cells

- Annarita Patrizi, PhD, Children's Hospital Boston

Rescuing Misregulation of NMDA Receptor Subunits in Rett Syndrome

"The possibility of new treatments for Rett syndrome increases as our research program broadens and intensifies," said Dr. Kaminsky. "I would like to thank our Board and our donors for their demonstrated commitment to advancing the best science in the community."

#### About Rett Syndrome

Rett syndrome (RTT), a neurological disorder, occurs almost exclusively in females. RTT results in severe movement and communication problems following apparently normal development for the first six to 18 months of life. Characteristic features of the disease include loss of speech and purposeful hand use, repetitive hand movements, abnormal walking, abnormal breathing, slowing in the rate of head growth and increased risk of seizures. Current treatment for girls with RTT includes physical and occupational therapy, speech therapy, and medication for seizures. There is no known cure for RTT. In 2007, researchers heralded a major breakthrough by reversing RTT symptoms in mouse models. RTT is considered a "Rosetta Stone" that is helping scientists understand multiple developmental neurological disorders, and shares genetic links with other conditions such as autism and schizophrenia.

#### About the International Rett Syndrome Foundation

IRSF is the world's leading private funder of basic, translational and clinical Rett syndrome research, funding \$30M in high-quality, peer-reviewed research grants and programs to date. Annually, IRSF hosts the world's largest gathering of global Rett researchers and clinicians to establish research direction and priorities while exchanging ideas and the most recent information. IRSF is the most comprehensive non-profit organization dedicated to providing thorough and accurate information about Rett syndrome, offering informational and emotional family support, and stimulating research aimed at accelerating treatments and a cure for Rett syndrome and related disorders. IRSF has earned Charity Navigator's most prestigious 4 star rating. To learn more about IRSF and Rett syndrome, visit [www.rettsyndrome.org](http://www.rettsyndrome.org) or call IRSF at 1-800-818-RETT (7388).