Press Release:
Treatment for Rett Syndrome Moves a Step Closer as RSRT Commits Record $4.2 Million in 2012

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The Rett Syndrome Research Trust (RSRT) affirmed its mission to drive innovative science by awarding $4.2 million in 2012. This amount represents the largest research commitment in a given year ever allocated by a Rett organization. The awards were made possible by the fundraising efforts of highly motivated families and supporters of children and adults with Rett Syndrome in the US, and through our global partnership with RSRT UK and Rett Syndrome Research & Treatment Foundation (Israel).

Rett Syndrome, the most physically disabling of the autism spectrum disorders, is caused by random mutations in the gene MECP2. Predominantly affecting girls, a frightening regression in toddlerhood robs children of the ability to speak, move normally and use their hands. Disordered breathing, Parkinsonian tremors, severe anxiety, seizures, digestive, circulatory and orthopedic problems typically appear. Although most children survive to adulthood they require total round-the-clock care.

“Our goal is not to fund research but to fund results. Ultimately, the measure of our success will not be in terms of dollars spent but whether we will have dramatically improved the quality of life of the people we serve – children and adults with Rett Syndrome. We believe the projects we chose to support in 2012 will help us achieve that goal,” said Tony Schoener, Chairman of the Board of RSRT.

RSRT awarded funds in 2012 to the following projects:

- **$2.2 million** was awarded to Benjamin Philpot at the University of North Carolina at Chapel Hill and his collaborators, Bryan Roth and Terry Magnusson. Their bold project is a screening initiative to identify compounds able to reactivate the silenced but healthy MECP2gene on the inactive X chromosome. RSRT has championed this approach since the game-changing discovery in 2007 that severe Rett-like symptoms in mice models can be reversed, even in late stages of the disease.Benjamin Philpot explains, “If a drug could be identified to efficiently and effectively activate MECP2, we would be attacking Rett at its very root, with the potential of reversing the disorder. Our entire team is excited about the possibilities and we’re ramping up the project as fast as possible.”

- **$760K** to Monica Justice’s lab at Baylor College of Medicine
  Funding of ongoing experiment to identify modifier genes – meaning alterations in genes that can dampen the ill effects of an MECP2 mutation. It was expected that modifier genes would be rare and difficult to find, but with the screen 15% complete, five modifiers belonging to diverse biochemical pathways have already been identified. It is likely that many more modifiers are waiting to be discovered. RSRT has therefore made finishing the screen a high priority, and this award brings our total commitment to the project to $1.5 million.

- **$720K** to Jonathan Kipnis at the University of Virginia
  This is additional funding to enable a better understanding of the immune system’s involvement in Rett. The proposed research would for the first time examine the role of the immune system in tissue from individuals with Rett Syndrome and could provide a foundation for potential immune-system therapies. Previous work from the Kipnis lab suggested that bone marrow transplants could provide some degree of symptomatic relief. Funding of $55K was awarded to independently confirm the Kipnis data.

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- **$240K** to Huda Zoghbi of Baylor College of Medicine
  Dr. Zoghbi's project seeks to determine whether symptoms of MECP2 Duplication Syndrome can be reversed once the protein is brought down to normal expression levels. These funds were awarded via the MECP2 Duplication Syndrome Fund at RSRT through the efforts of the duplication/triplication families.

- **$65K** to Sasha Djukic
  Support for continuing work at the Tri-State Rett Syndrome Center in the Bronx, NY with the majority of the funds raised through the annual Reverse Rett NYC event.

- **$175K** to the labs of John Bissonnette at OHSU and Andrew Pieper at UTSW for drug repurposing explorations.

Monica Coenraads, RSRT's Executive Director, said: "Our approach to funding research is somewhat different from that of more traditional research organizations. We don't request proposals and wait, hoping for someone to submit an interesting project. Instead, we spur the research agenda by engaging closely with the scientific community each step of the way, so that together we can identify the most promising areas and invest in them quickly. As parents of daughters with Rett Syndrome, we don't have a moment to lose."

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rett syndrome research trust

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.