rett syndrome research trust

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RSRT Key Facts

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RSRT Key Facts

WHO WE ARE

The Rett Syndrome Research Trust (RSRT) is a non-profit organization with a highly personal and urgent mission: a cure for Rett Syndrome and related MECP2 disorders.

RSRT is led by:

- *Eleven-member Board of Trustees (eight are parents of daughters with Rett; one has a sister with Rett; one is a scientist; one is a friend of an affected family)*
- *Five full time staff (Executive Director and Program Director have daughters with Rett)*
- *Thirteen-member Scientific Advisory Board includes some of the most respected scientists in the world*
- *Co-founder and Executive Director, Monica Coenraads, has been the one constant for the Rett research community since 1999*

OUR HISTORY

Monica became involved in Rett research in 1998 the day her then two-year-old daughter was diagnosed. She established the Rett Syndrome Research Foundation (RSRF) a year later with five other parents and served as Scientific Director. In 2007 RSRF merged with the older, family support-focused International Rett Syndrome Association (IRSA), founded by Kathy Hunter in 1984. The combined entity was renamed the International Rett Syndrome Foundation (IRSF), now doing business as RettSyndrome.org . Monica, eager to focus strictly on research, resigned and launched RSRT in 2008 with Ingrid Harding.

RSRT Key Facts

WHAT WE DO

RSRT operates at the nexus of scientific activity. We enable advances in knowledge and drive innovative research through constant engagement with academic scientists, clinicians, industry, investors and affected families. These relationships catalyze the development and execution of a research agenda that neither academia nor industry could achieve alone.

We identify, solicit, evaluate, prioritize, fund and monitor ambitious research projects that have the greatest likelihood of impacting the lives of those afflicted with Rett Syndrome.

We have provided financial support to:

- *44 scientists in 36 academic institutions*
- *4 biotech companies*
- *5 clinical trials testing 4 compounds*
- *Conceptualized and launched 4 consortia (MECP2 Consortium, Gene Therapy Consortium, Activating the Silent MECP2, Outcome Measures and Biomarker Development))*

YEAR	$ AWARDED TO RESEARCH	% OF DONATIONS TO RESEARCH
2008	2.3	98%
2009	0.6	94%
2010	1.3	96%
2011	3.6	96%
2012	4.3	97%
2013	7.2	96%
2014	5.8	93%
2015	8.8	94%
2016	2.0 (YTD)	TBD
TOTAL	$36 MILLION	96% (AVERAGE)

RSRT Key Facts

WHAT WE DO *(Continued)*

We refute the conventional practice of labs working in isolation and promote and fund collaborations and consortia in which scientists work across multiple disciplines. We are risk takers who do not shy away from bold projects that are unlikely to be funded by NIH or other more traditional funding agencies.

Through close monitoring of scientific literature we identify and recruit the interest of scientists doing work relevant to our mission. We organize scientific meetings throughout the year favoring small, invitation-only gatherings to promote in-depth discussion and ensure participants of confidentiality, allowing them to share data long before publication, a process that can take years.

We provide accurate, timely research information to the Rett family community without bias or spin. We operate with transparency and strict accountability to the public and to Rett families in particular. We are funded almost entirely by parents of children with Rett and their networks in the US and abroad.

OUR ACCOMPLISHMENTS

Basic Scientific Discoveries- Expand our understanding of Rett and MECP2

- *Discovered that the Rett protein, MeCP2, is required throughout life to maintain healthy brain function. (Adrian Bird, Huda Zoghbi)*
- *Discovered that the genes disrupted in Rett are much longer (over 100,000 nucleotide bases) that the average length of a gene (20,000 nucleotide bases). Labs now testing drugs that specifically down regulate long genes, such as topoisomerase inhibitors. (Michael Greenberg via MECP2 Consortium)*
- *Identification of a critical novel domain on the Rett protein (NCoR/SMRT Interaction Domain) that could have therapeutic implications for Rett and for MECP2 Duplication Syndrome. (Adrian Bird via MECP2 Consortium)*
- *The creation and distribution of a number of clinically relevant Rett mouse models that mimic the human disease. These mice provide a useful basis for pre-clinical tests of therapeutic approaches. (Adrian Bird and Michael Greenberg via MECP2 Consortium)*
- *The revelation that glia play a key role in preventing the progression of the most prominent Rett symptoms displayed by mouse models of the disease.*

RSRT Key Facts

OUR ACCOMPLISHMENTS *(Continued)*

Translational Discoveries – Could lead to new cures and therapies

- *Dramatic symptom reversal in Rett mice using gene therapy treatment. (Gail Mandel & Adrian Bird)*
- *Discovered that the MECP2 Duplication Syndrome is reversible. (Huda Zoghbi)*
- *Established two entirely novel areas of Rett research:*
  - *1) Activating the Silent MECP2 Gene*
  - *2) Modifier Genes*
- *Deep Brain Stimulation may be helpful for Rett and the MECP2 Duplication Syndromes.*
- *Funded 4 clinical trials (ketamine, copaxone, statin, triheptanoin) for symptom improvement.*

Enabling Technology – Tools and processes that are necessary to accelerate discovery and development of cures and treatments

- *Conceived of and launched the Gene Therapy Consortium to advance gene therapy treatment to clinical trials ASAP.*
- *Conceived of and launched the Outcome Measures and Biomarker Development Consortium, a $4.5 million project to identify and validate FDA accepted endpoints for clinical trials in Rett.*
- *Organized scientific meetings and workshops every year, throughout the year that include the pharmaceutical industry and investors to increase their interest in Rett Syndrome.*

Organizational Accomplishments

- *RSRT is the only Rett organization in the world that is aggressively funding multiple approaches that could lead to a cure: gene therapy, protein replacement, activating the silent MECP2.*
- *Since 2008, RSRT has awarded $36 million to research, more than any other Rett organization*
- *RSRT is lean. Since our launch an average of 96% of every dollar donated has been channeled to our research program.*
- *Cultivated global partnerships with Reverse Rett (UK), Rett Syndrome Research & Treatment Foundation (Israel), Rett Syndrome Deutschland, Rett Syndrome Ireland, and more.*
- *Cultivated national partnerships with Girl Power 2 Cure, Rocky Mountain Rett Association, Rett Syndrome Association of Massachusetts, Rocky Mountain Rett Syndrome Association, and more.*
- *We refute the conventional practice of labs working in isolation, instead we promote and fund consortia and collaborations in which scientists work across multiple disciplines.*