

Leadership	P. 1
Research	P. 3
Family Support	P. 7
Fundraising	P. 15
Our Heroes	P. 19

Rett Gazette

FALL • 2013

INTERNATIONAL RETT SYNDROME FOUNDATION



IRSF
INTERNATIONAL
RETT SYNDROME
FOUNDATION

PRO RETT RICERCA SUPPORTS THE IRSF SCOUT PROGRAM WITH A \$200,000 GIFT

The Italian Rett syndrome association Pro Rett Ricerca has partnered with the International Rett Syndrome Foundation to launch IRSF's latest initiative, the IRSF Scout Program. The generous \$200,000 commitment from Pro Rett Ricerca will allow IRSF to test 8 of the 24 compounds to be tested in this ground-breaking program to identify possible treatments for Rett syndrome.

*Rita Bernardelli, President
of Pro Rett Ricerca
says about the partnership:*



"We are convinced that collaboration between associations and foundations from different countries is necessary to speed up a cure for girls with Rett Syndrome, each of them dedicating a part of their funds towards common research projects requiring a large scientific and financial effort.

Large foundations such as IRSF have the resources to develop ambitious scientific projects with excellent scientists who guarantee research of good quality; they define goals and different procedures. They are also able to find research companies who can help in developing the projects faster compared to the smaller labs.

This is why Pro Rett Ricerca has decided to support the Scout Program which is very ambitious and we would never have been able to do it on our own, but since our same goals in finding a cure are urgent and in common with those of IRSF we have chosen to support part of this project.

We invite other family associations around the world to follow our example, because together we can reach our goals faster."



Left to right: Rita Bernardelli, president, Roberto Tisato, board member and Laura Rasetti, vice president

IRSF celebrates this new alliance, which will allow us to make a great step forward in our search for a safe and effective medicine for Rett syndrome:

"When developing a treatment for Rett syndrome, we are looking at a hallway with many doors. To open the door to see what's in that room costs us \$25,000 per door. Pro Rett Ricerca is helping us to open 8 of those doors," said Dr. Steven Kaminsky, IRSF Chief Science Officer, who has developed the innovative Scout Program with the goal to speed up the development of an effective treatment for Rett syndrome.

Pro Rett Ricerca is an Italian association founded in 2004 by a group of parents of children with Rett syndrome. Their mission is to support medical research with the ultimate goal to develop a treatment and a cure for Rett syndrome. In 2011, Pro Rett Ricerca established the San Raffaele Rett Research Center, the first Italian research laboratory dedicated solely to Rett syndrome. The work of Pro Rett Ricerca is directed by their vision that synergic collaboration among organizations dealing with Rett syndrome is crucial to reaching the common goal of finding a treatment and a cure for children living with Rett syndrome as soon as possible. To learn more about Pro Rett Ricerca, please visit their website at www.prorett.org.

The Rett Gazette is published periodically by the International Rett Syndrome Foundation (IRSF), a non-profit 501(c)(3) organization. The core mission of the IRSF is to fund research for treatments and a cure for Rett syndrome while enhancing the overall quality of life for those living with Rett syndrome by providing information, programs, and services.

BOARD OF DIRECTORS:

Dr. Rick Altschuler
Daniel Brinkhaus
John Corpus, Treasurer
Eric Diamond
Pamela Diener
Bill Farnum
John Foard
Stephen Gallucci
Steven Jones
Michael Joyce, Vice Chair
Kathryn Kissam
Barry Rinehart
Judi Rees
Kenna Seiler, Secretary
Rajat Shah, Chairman

EDITOR:

Jennifer Endres

ABOUT RETT SYNDROME

Rett syndrome (RTT) is a devastating postnatal neurological disorder that occurs almost exclusively in females. It is usually caused by a mutation of the MECP2 gene on the X chromosome. Rett syndrome is found in all racial and ethnic groups throughout the world, and in every socio-economic class. RTT affects 1 in 10,000 live female births. Early developmental milestones appear normal, but between 6–18 months of age, there is a delay or regression in development, particularly affecting speech, hand skills, and coordination. A hallmark of Rett syndrome is repetitive hand movements that may become almost constant while awake. Other features may include seizures, irregular breathing, swallowing difficulties, deceleration of head circumference with age, and curvature of the spine. Many individuals with Rett syndrome live well into adulthood. There is currently no cure.

All material in the IRSF newsletter is copyrighted and most may be reproduced with permission. Call 1-800-818-RETT (7388). Back issues are available for \$2 per copy. All submissions and correspondence regarding the newsletter, information and partnerships should be directed to:

IRSF

4600 Devitt Drive, Cincinnati, OH 45246
1.800.818.RETT (7388) or 513.874.3020
admin@rettsyndrome.org
Fax Number: 513.874.2520
EIN Number: 31-1682518

POLICY STATEMENT

In an effort to improve the quality of life for those with Rett syndrome we welcome ideas from our readers on various techniques of management and care, general and educational suggestions that individuals have investigated and/or found beneficial. Parents and others have the opportunity to decide if they want to follow through with the suggestions. As a foundation, we do not support or endorse any particular treatment, therapy, or medication. We encourage parents to support one another with suggestions and to contact their child's physician for final approval.

© 2013 IRSF. All rights reserved.

COMING IN 2014...

RETT SYNDROME HANDBOOK ON RETTSYNDROME.ORG!



YES, THAT'S RIGHT! Leave that 4 pound book of gold at home but don't leave home without the Info! Coming in 2014 for your smart phone and tablet. Users will have the valuable information, wisdom and trusted advice from the internationally recognized Rett Syndrome Handbook at your fingertips. Functionality will be added to content to make it a powerful, customized, personal tool for educating others about Rett syndrome, as well as YOUR child. Rich media, new images, videos, and ability to track your child's development.

WHAT A CONCEPT! This represents a major breakthrough for empowering families, brought to you by your trusted friends at IRSF. It will be free, and it will be available to all worldwide.



Message from IRSF's Chief Science Officer
Dr. Steven G. Kaminsky

Greetings

As 2013 closes, it is a good time to slow down a little and look at the research accomplishments as we get ready for our next year. The year started out great with the first gene therapy publication by the laboratories of Stuart Cobb and Steven Gray that showed that gene therapy is a viable approach to treating Rett syndrome. Next Monica Justice's lab published work to show that modifying the cholesterol metabolism should be considered as a new avenue to change the biology of Rett. More recently, an exciting discovery came out of MIT using the latest of genetic technologies (TALEN) changing our fundamental views of Rett molecular biology. During all of this, the two clinical trials that IRSF is sponsoring in Boston and Houston kept moving forward at their anticipated rate, and we all anxiously await the final evaluation of these trials in mid 2014. Finally, IRSF started a new program, the Scout Program, to bring more potential therapies forward for clinical trials at an accelerated rate in the near future.

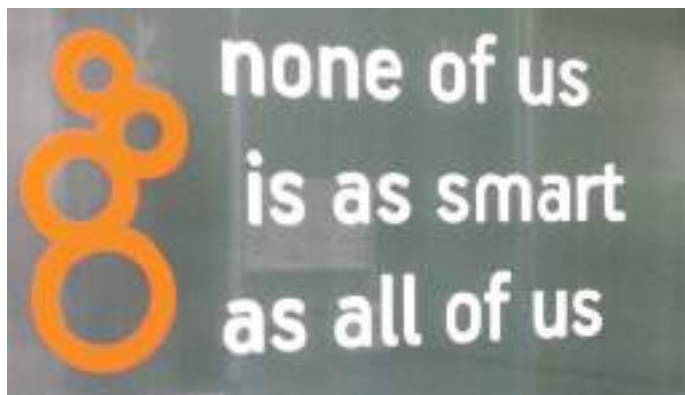
All of this, along with a very successful Family Conference in Midway, Utah, paints a very good picture for 2013. But we still have more to do! With each discovery, our mission to identify possible treatments for Rett syndrome gets closer. With each discovery, our excitement grows! However, none of this would be possible without the tremendous sacrifices each of you have made to raise money for research into this disorder. My inspiration is found in each of you. As I get older, I have come to realize that it takes a team to get most things done. Rarely do we do anything on our own. At Midway, Utah we, as a community, felt wildly optimistic about the outlook for the near future and the possible treatments on the horizon.

Let's keep the momentum that was started at that Family Conference and recruit more individuals to our cause of helping each of our girls and boys with Rett syndrome and their families. As we look at what we are all collectively doing, none of us can do everything that needs to be done. But each of us can do something that makes a difference. So please continue what you are doing and help make our dream of identifying treatments happen. We are strongest when we play as a team knowing that our final goal is to treat Rett syndrome and improve the quality of life for each of our girls and boys and their families affected by this disorder.

Sincerely,
Steve Kaminsky, Ph.D.

2013 IRSF Research Meeting News

This past October, IRSF was a sponsor of the 3rd European Conference in Maastricht, The Netherlands. Because of this big meeting, we decided not to put together the usual Annual IRSF Research Symposium this year. We opted to hold Program Reviews in San Diego, CA and Boston, MA. These two locations were chosen based on the number of Rett Researchers (not just IRSF funded ones) in those areas. IRSF funded investigators were asked to present their current progress. We heard about all areas of research including basic discovery, translational, and clinical research by these investigators in San Diego and Boston. At each meeting, we were impressed about the work being done, the collaborations being made, and the passion these investigators radiated. These visits were well received, and the format of the meeting was intimate and informal bringing ease to open discussion of non-published and preliminary work. The motto on the walls of the Sanford Consortium where we held the meeting in San Diego summed up the purpose for our program meetings perfectly, "None of us is as smart as all of us." Let's continue to work together and chart the course to treat Rett syndrome.



Planning for the 2014 Research Meeting is Underway

In June 2014 we will host the IRSF Research Symposium in the DC area. IRSF CSO Steve Kaminsky has put together the planning committee and together, they are outlining four exciting sessions. The planning committee includes the following chairs: Dr. Alan Percy – Clinical Advances; Dr. David Katz – Translational Advances and Challenges; Dr. Sacha Nelson – Basic Discovery Updates; and Dr. Pamela Diener – Neuro-Abilitation, sensory-motor education. Please be on the lookout for upcoming information!

Research Funding News

Scout Program: Accelerating the Pace of Translational Research for Rett Syndrome.

The International Rett Syndrome Foundation has decided to pave a new path for Rett syndrome research. Our mission is to move discoveries to the clinic. We believe that the Scout Program will accelerate the testing of compounds, whether new or repurposed, through standardized preclinical studies in the Rett mouse model. By using a contract research organization, we will ensure the studies will yield robust and reproducible results. It will help identify compounds that have potential versus those that will fail. We want those that fail to “fail fast” so we can put more effort into those with potential. In June 2013, the IRSF Board of Directors gave final approval to fund \$600,000 towards the Scout Program. This program will test 20-24 compounds over the next two years.

A team of seven scientists will serve on the Scout Advisory Committee for the Scout Program and together, they will prioritize the biological pathways to target and the compounds to be tested.

- Dr. John McCall, PharMac, LLC
- Dr. David Katz, Case Western Reserve University
- Dr. Garry Smith, Fox Chase Chemical Diversity Center, Inc.
- Dr. Stephen Ginsberg, Nathan Kline Institute, New York University
- Dr. Aleksandra Djukic, Montefiore Medical Center, Albert Einstein College of Medicine
- Dr. Rajiv Ratan, Burke Medical Research Institute, Cornell University
- Dr. Marcie Glicksman, Harvard University

IRSF has executed a contract with the Contract Research Organization (CRO) PsychoGenics, Inc. in Tarrytown, NY. Dr. Daniela Brunner, Senior Vice

President of Behavioral R&D will be leading the testing team.

We are now at the point of working with our Advisory Committee and prioritizing the compounds.

PsychoGenics is ready to move forward once the compounds are available. We are anticipating that the first round of compound testing will be complete by the first quarter of 2014.

Continuing our push for more Clinical Research

Now with the implementation of the new Scout Program and the two IRSF supported Phase 2 clinical trials underway, we eagerly anticipate a lot of activity on the clinical research front in the near future. We want to be ready to move this research momentum further. With this in mind, the IRSF Board has approved an additional \$0.5 M in anticipation of future critical trials. It is quite possible that the current compounds will move to a Phase 3 study as long as they yield positive efficacy results in Phase 2. In addition, compounds may move quickly through the Scout Program into a Phase I clinical trial upon FDA approval. Either way, we will be ready to push compounds through the drug discovery pipeline to the clinical side.

New Translational Research Grants Awarded in June 2013

Last year, the IRSF Board of Directors had implemented a change to the early stage Translational Research grant mechanism called Help Accelerate Rett Therapeutics (HeART) to enable research investigators to better facilitate the development and testing of novel therapeutics. The restructured HeART gives them more time and more resources for additional staff and supplies to conduct the experiments. The HeART award now has a maximum funding of \$150,000 to be spent over a 2 year period of time (\$75K total costs/year).

IRSF just completed the first cycle of the IRSF Translational Research Grant

Program using this updated mechanism. A peer review of 17 applications was conducted during the spring, and the IRSF Board approved the funding for two of the HeART applications.

2013 HeART Awards



Theresa E Bartolotta, PhD



Patricia Remshifski, PhD

Theresa E Bartolotta, PhD, Seton Hall University and Patricia Remshifski, PhD, Monmouth University
\$60,000

“Eyetracking in Rett syndrome: A preliminary investigation of receptive and expressive vocabulary”



John Christodoulou, AM

John Christodoulou, AM, Sydney Children's Hospital Network, Westmead, Australia
\$150,000

“Preclinical Evaluation of Tubastatin A, a Novel Therapy for Rett Syndrome”

**Heartfelt thanks
from IRSF to all
of our supporters.
Your generosity has
allowed us to launch
this groundbreaking
Scout Program.**

MEET DANIELA BRUNNER

Now that IRSF has announced the Scout Program, we wanted you to meet the person overseeing the drug discovery screen in the animal model– Daniela (Dani) Brunner, PhD, Senior Vice President of Behavioral Research and Development at PsychoGenics, Inc. Dr. Brunner has published extensively in behavioral pharmacology, animal models of disease, mathematical models of behavior, and has written several book chapters on such subjects.

Dr. Dani Brunner grew up in Argentina. She always had an interest in science, animal behavior in particular, following influences from her father, a photographer and scuba diver, and her mother, a mathematician and publisher. Following her interests in nature, she became a biologist and majored in Zoology at Buenos Aires University. At the end of 1989, Dr. Brunner earned a PhD in Experimental Psychology in Cambridge University, England, where the focus of her studies was the mathematical modeling of optimal behavior (i.e. the study of decision making in the wild). Dr. Brunner then moved to the United States where she completed postdoctoral training at Columbia University, studying decision making in normal animals first, and then in animal models of disease. Such animal models harbored genetic mutations that affected both development and adult behavior, and thus she expanded her interests to developmental psychobiology and psychiatry. After her postdoc, Dr. Brunner moved to Hunter College, City University of New York (CUNY), where she taught statistics and experimental design. She remains associated with Columbia University as a faculty member collaborating in research and teaching from time to time.

Dr. Brunner was one of the first employees of PsychoGenics, as Director of Behavioral Research in 1999, when she set up a large battery of standard behavioral tests and soon thereafter started the development of several proprietary in vivo platforms, for which she holds several patents. She expanded their animal colonies and was responsible for bringing the Bird Rett mouse model to the company. It was an exciting time to begin a more extensive analysis of these mice. The groundbreaking study of the reversal of Rett syndrome in this animal model had just been published two years earlier in 1997. Dr. Brunner wanted to understand these mice and knew it was essential to study their behavior and determine how close they modeled the human disorder. In 2011, Dr. Brunner presented this work at the NINDS



*Daniela (Dani) Brunner, PhD
Senior Vice President of
Behavioral Research
and Development at
PsychoGenics, Inc.*

workshop on “Setting Standards for Preclinical Research for Rett Syndrome”. An intricate and detailed description of their social behavior, motor skills, and gait analysis was captured using PsychoGenics’ highly advanced technology. It is this technology that will be used in the Scout Program of in vivo drug screening and will quickly determine which compounds have therapeutic potential. All proposed testing will follow a standardized preclinical protocol aligned with the recommendations of the workshop.

Dani Brunner stated that “working in science is rewarding, but when you know your work will directly help to relieve the disease burden of affected kids and their families, then it becomes truly rewarding. We are very happy to be able to work closely with the foundation.”

PsychoGenics is located in Tarrytown, NY. As a contract research organization and drug discovery company, it offers a wide range of preclinical services and has developed partnerships with both disease-oriented nonprofit organizations as well as other corporate institutions. The President and CEO of PsychoGenics, Dr. Emer Leahy, is also a member of the IRSF Scientific Advisory Board and has intimate knowledge of our mission.

We are thankful to have Dr. Brunner and her team at PsychoGenics to move compounds through standardized preclinical testing in the Rett animal model. Our hope is that the compounds that pass her robust testing will be accelerated in their path to future clinical trials and bring us closer to new treatments for those diagnosed with Rett syndrome.

New Inclusion Criteria for the IGF-1 Clinical Trial in Boston were announced in October.

1. The participant age range is now 2-10 years for those past the regression stage.
2. The participant can reside in the USA or Canada.

See <http://www.rettsyndrome.org/research-programs/clinical-trials-and-databases/rett-syndrome-specific-trials#IGF1> for more details.

What are Fast Track Status and Breakthrough Therapy Status at the U.S. Food and Drug Administration?

With the International Rett Syndrome Foundation's focus on pre-clinical and clinical trials, many have asked the question about United States Food and Drug Administration's (FDA) designators of "Fast Track Development Program" and "Breakthrough Therapy Program". Much of what is written below is taken directly from the FDA website in regard to their programs.

The Fast Track Development Program is designed to accelerate the approval of investigational new drugs getting ready for clinical trials with the goal review time of 60 days. Such status is often given to agents that show promise in treating serious, life-threatening medical conditions for which no other drug either exists or works as well. The purpose is to get important new drugs to the patient earlier. Fast Track addresses a broad range of serious diseases. Determining whether a disease is serious is a matter of judgment, but generally is based on whether the drug will have an impact on such factors as survival, day-to-day functioning, or the likelihood that the disease, if left untreated, will progress from a less severe condition to a more serious one. Any drug being developed as a treatment or preventative measure for a disease that does not have a current therapy is labeled as an unmet need. If there are existing therapies, a fast track drug must show some advantage over available treatment, such as:

- Showing superior effectiveness
- Avoiding serious side effects of an available treatment
- Improving the diagnosis of a serious disease where early diagnosis results in an improved outcome
- Decreasing a clinically significant toxicity of an accepted treatment

A drug that receives Fast Track designation is eligible for some or all of the following:

- More frequent meetings with FDA to discuss the drug's development plan and ensure collection of appropriate data needed to support drug approval
- More frequent written correspondence from FDA about such things as the design of the proposed clinical trials

- Eligibility for FDA Accelerated Approval, i.e., approval on an effect on a surrogate, or substitute endpoint reasonably likely to predict clinical benefit
- Rolling Review, which means that a drug company can submit completed sections of its New Drug Application (NDA) for review by FDA, rather than waiting until every section of the application is completed before the entire application can be reviewed. NDA review usually does not begin until the drug company has submitted the entire application to the FDA
- Dispute resolution if the drug company is not satisfied with an FDA decision not to grant Fast Track status.

Fast Track designation must be requested by the drug company. The request can be initiated at any time during the drug development process. FDA will review the request and make a decision within sixty days based on whether the drug fills an unmet medical need in a serious disease.

Once a drug receives Fast Track designation, early and frequent communication between the FDA and a drug company is encouraged throughout the entire drug development and review process. The frequency of communication assures that questions and issues are resolved quickly, often leading to earlier drug approval and access by patients.

On July 9, 2012 the Food and Drug Administration Safety and Innovation Act (FDASIA) was signed. FDASIA Section 902 provides for a new designation - **Breakthrough Therapy Designation**. A breakthrough therapy is a drug:

- Intended alone or in combination with one or more other drugs to treat a serious or life threatening disease or condition and
- Preliminary clinical evidence indicates that the drug may demonstrate substantial improvement over existing therapies on one or more clinically significant endpoints, such as substantial treatment effects observed early in clinical development.

A drug that receives Breakthrough Therapy designation is eligible for the following:

- All Fast Track designation features
- Intensive guidance on an efficient drug development program, beginning as early as Phase 1
- Organizational commitment involving senior managers

If a drug is designated as breakthrough therapy, FDA will expedite the development and review of such drug. All requests for



BLUE BIRD CIRCLE TEAM

*L to R – First Row: Lata Casturi, Dr. Daniel Glaze, Jacki Soto, Thuy Dinh C-PA, Rory Mahabir
Second Row: Robert McNeil, Amber Pearce, Dr. Jeff Neul, Aryn Knight, Wes Moulden, Judy Barrish, Jason Vecchio*

breakthrough therapy designation will be reviewed within 60 days of receipt, and FDA will either grant or deny the request.

So what are the differences between the breakthrough therapy designation and the fast track designation?

Although breakthrough therapy and fast track designation programs have many similarities, as they both are intended to expedite the development and review of drugs for serious or life-threatening conditions, there are differences in what needs to be demonstrated to qualify for the programs. A breakthrough therapy program is for a drug that treats a serious or life-threatening condition and preliminary clinical evidence indicates that the drug may demonstrate substantial improvement on a clinically significant endpoint(s) over available therapies. In contrast, a fast track program is for a drug that treats a serious or life-threatening condition, and preclinical data demonstrate the potential to address unmet medical need.

Fast Track and Breakthrough Therapy are just a couple approaches of the FDA that are intended to make therapeutically important drugs available at an earlier time. It is important to note that standards for the safety and effectiveness of such drugs that become available through these processes are not compromised. NNZ-2566 is one of the drugs IRSF is supporting that has been given the Fast Track Status, and Neuren Pharmaceuticals continue to have an open dialogue with those at the FDA. We hope that more compounds that come to the end of the drug discovery pipeline will have enough preclinical and/or clinical evidence to be given either of these designations. IRSF will continue to participate in such discussions with the academic labs, biotech and pharmaceutical companies, and the FDA to expedite drug development and review for Rett syndrome.

IMPORTANT NATURAL HISTORY STUDY UPDATE

Changes to Travel Sites (CA, FL, IL, NJ) and New Grant Application

Alan K. Percy, MD, Principal Investigator, Rett Syndrome Natural History Study



The Rett syndrome (RTT) project involves the largest population under study with classic and variant RTT and, including individuals with related disorders, now represents an enrollment of 1170. Individuals with classic and variant RTT began to be enrolled in this study in 2006 and were followed at least annually. Current enrollment is almost completely female and involves nearly 1000 individuals with typical or atypical RTT. During the same period, nearly 100 individuals were enrolled that 1) have a mutation in MECP2, but do not fulfill criteria for RTT, 2) have MECP2 duplications, or have mutations in CDKL5. We have continued to enroll new participants at the four investigative sites, Children's Hospital Boston (Harvard), Baylor College of Medicine, Greenwood Genetic Center, and the University of Alabama at Birmingham and at the four regional travel clinics visited twice yearly, a) Oakland Children's Hospital, b) Florida (once each annually to Miami and Tampa), c) Rutgers-University of Medicine and Dentistry of New Jersey, and d) Rush Medical Center in Chicago.

The current funding for this grant ends on July 31, 2014. We have recently submitted a renewal application for continued funding of these disorders including MECP2 duplications, CDKL5, FOXP1, and those with a MECP2 mutation but not meeting criteria for RTT. No assurance can be given that this grant will be funded, but in either case, the travel clinics as currently conducted will end at that time.

During the past several years, RTT clinics have developed across the country such that the travelling clinic component can be replaced by these regional clinics to continue the enrollment and follow-up of individuals with RTT and the

other RTT-related disorders. In this manner we will ensure broad geographic representation of clinical expertise to ease the travel issues for families or principal caregivers at the same time increasing the numbers of qualified clinical investigators available to conduct these critical assessments. These sites may include: UC San Diego (Dr. Richard Haas), Oakland Children's (Dr. Mary Jones), Rush Medical Center (Dr. Peter Heydemann), University of Colorado (Dr. Timothy Benke), University of Rochester (Dr. Alex Paciorkowski), Children's Hospital of Philadelphia (Dr. Eric Marsh), and the four original participating sites. Dr. Sarika Peters at Vanderbilt will be involved with the MECP2 duplication study.

In addition to the longstanding interaction and support from the International Rett Syndrome Foundation (IRSF), we have discussed and gained enthusiastic support for expansion of enrollment of this consortium from the three other patient advocacy groups: the MECP2 Duplication Foundation, the International Foundation for CDKL5, and the International FOXP1 Foundation to include individuals with these three disorders. They will serve important roles in recruitment and retention of research participants and will also provide for the exchange of critical information between the parents or other caregivers and the research teams.

We appreciate most sincerely the many families who have participated in the travel clinics. We have made great progress that would not have been possible without this effort by you. The regional clinics will continue this research effort with the commonly held goal of reversing the impact of these neurodevelopmental disorders.

Nutritional and Digestive Health Booklet - Now Available!

An information booklet for improving health outcomes in Rett syndrome has been produced by the Rett syndrome study team at the Telethon Institute for Child Health Research, Perth, Western Australia.

The aim is to provide a ready reference for families and caregivers on the best ways of improving the nutritional and digestive health of girls and women with Rett syndrome.

Published by the same team who brought you the Scoliosis Management Guidelines funded in part by IRSF.



We encourage all families to visit and download this new care guide: <http://rett.childhealthresearch.org.au/resources/guidelines,-reports-and-books.aspx>

A PARTICIPANT'S GUIDE TO DRUG RESEARCH IN RETT SYNDROME



IRSF has created "A Participant's Guide to Drug Research in Rett Syndrome" that aims to help parents or legal guardians of a child with Rett syndrome understand the importance of clinical trials and what participation means, what to expect, and what your rights are as well as the rights of your participating child.

World Rare Disease Day February 28, 2014

Rare Disease Day is an international advocacy day to bring widespread recognition of rare diseases as a global health challenge. It has grown significantly as a global event, and as a partner in this awareness day, we encourage everyone to plan something to raise awareness about Rett syndrome as a Rare Disease on this critical day. No effort is too small. Media attention, or community awareness, it all matters in our goal to find partners and support for research, advocacy, and awareness.



In the U.S., any disease affecting fewer than 200,000 people is considered rare. This definition comes from the Orphan Drug Act of 1983 and is slightly different from the definition used in Europe. There are nearly 7,000 rare diseases affecting nearly 30 million Americans. In other words, almost one in ten Americans are suffering from rare diseases.

Alone We Are Rare, Together We Are Strong

rarediseaseday.us



You can download this Guide from the IRSF website page called Family Resource Center (<http://www.rettsyndrome.org/news-and-media/family-resource-center>).

CLINIC SPOTLIGHT

NEW Rett Syndrome Clinic at Cincinnati Children's Hospital Medical Center

Drs. Shannon Standridge and Patty Manning
Pediatric Neurology and Developmental and Behavioral Pediatrics
Co-Directors, Rett Syndrome Clinic

Cincinnati Children's Hospital Medical Center
3333 Burnet Avenue
Cincinnati, OH 45229

Contact: Anita Boyer, Administrative Coordinator, 513-636-9214
rett@cchmc.org
For appointments call: 513-636-4222 option 1

DESCRIPTION OF CLINIC

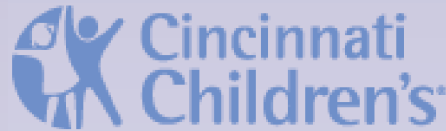
The Rett Syndrome Clinic at Cincinnati Children's Hospital Medical Center opened its doors in July, 2013. The clinic is housed within the Neurology division in building A, eighth floor. The clinic is multi-disciplinary and currently involves a core group of disciplines (neurology, developmental pediatrics, social work, nutrition, OT, and ST) in a full day clinic once a month. In order to care for other important medical concerns in patients with Rett Syndrome, care management relationships with other medical providers such as orthopedics, pulmonary, gastroenterology, and gynecology are established or being developed to help families navigate this care.

CLINIC SCHEDULE

Full day the fourth Thursday of each month.

CLINIC LOCATION

Neurology clinic space building A, eighth floor
Cincinnati Children's Hospital Medical Center
MLC 2015, 3333 Burnet Avenue
Cincinnati, OH 45229



FUTURE COLLABORATION OPPORTUNITIES

A number of RTT clinics have been started across the US, and Cincinnati Children's Hospital Medical Center is well positioned to contribute to the important work that has already started by collaborating and becoming a leader in research efforts

BIOSKETCH OF PROVIDERS

SHANNON STANDRIDGE, DO, MPH

Dr. Shannon Standridge completed her general pediatrics and neurology residency at Nationwide Children's Hospital. She then completed an electrophysiology fellowship at Cincinnati Children's Hospital Medical Center. Currently, she is a pediatric neurologist who specializes in treating epilepsy at Cincinnati Children's Hospital Medical Center. In her career, Dr. Standridge has developed a strong interest in treating children with Rett syndrome which has led to the opportunity to serve as a Co-Director of the Rett Clinic. Although she practices general neurology, she has specialized training in epilepsy management and she looks forward to focusing her career in the care of patients with Rett Syndrome.

PATTY MANNING, MD

Dr. Manning completed her residency in general pediatrics, as well as her fellowship in Developmental/Behavioral Pediatrics at Cincinnati Children's Hospital Medical Center. During fellowship, Dr. Manning developed an interest in children with autism spectrum disorders, including Rett Syndrome. Following fellowship, Dr. Manning launched The Kelly O'Leary Center for Autism Spectrum Disorders (TKOC), a multidisciplinary diagnostic, treatment and research program for children with autism spectrum disorders, including Rett Syndrome. Dr. Manning has maintained a strong interest in Rett Syndrome, and it has been her hope to be part of a multidisciplinary clinic for children with Rett syndrome. She has followed many girls with Rett Syndrome throughout their lifespan, and helped author a medical guideline for girls with Rett Syndrome.

SAVE THE DATES • SAVE THE DATES • SAVE THE DATES

Opening Doors to *DISCOVERIES*

ANNUAL FAMILY CONFERENCE
AND RESEARCH MEETING

National Capital Area

IRSF Basic, Translational, Clinical,
and Neuro-Abilitation Research Symposium
June 24-26, 2014

IRSF Family Education and Awareness Conference
June 26-28, 2014

Visit rettsyndrome.org
for registration and meeting details

POSSIBILITIES



Nora plays with her chicks



Sorel takes part in the Special Olympics



Mackenzie participates in school concert



Daphni has fun on the ferris wheel



Victoria makes her first communion



Cammy rides in a hot air balloon



Keira is a great baker



Rachael enjoys some time on the golf course



Sejal has a blast skiing

Just because our children have the diagnosis of Rett syndrome, it does not mean they cannot do everything their siblings and peers do. With a little effort and planning ahead as well as some patience and flexibility, your child can take part in a wide array of activities while remaining safe and sound. Participating feels good for the whole family but most importantly, it provides self-esteem to your special and unique child.



GRANDPARENTS

Be sure you have registered for the
Grandparents Network
[http://www.rettsyndrome.org/family-support/
support/online-support-networks](http://www.rettsyndrome.org/family-support/support/online-support-networks)
Join hundreds of others as you unite
to support each other, support your grandchild
and become a part of the Rett syndrome community.
Here are just a few of our special
grandparents with their granddaughters!

ABRIELLA AND TERRI

I love Abriella because she has opened my eyes so much to the world of RETT SYNDROME and the world of the disabled. She has enriched my life so much-I really feel like I'm here to help her and her family along this hard arduous path. And her laughter is so contagious! Just love her and all of the successes she has had! She just started walking a week before her 2nd b-day and is continuing to do really well although she has lost the functional use of her hands and speech! She is definitely a RETT ANGEL!



RILEY AND PAPA

We were all together at Camp New Hope, NC. It was a beautiful & relaxing week and we just had the best time with our granddaughter and her parents.



ISABELLA AND PA

God must have loved Isabella and us very much to have given her to us. I call this picture "LOVE". Love bursts forth as soon as anyone meets her. The picture couldn't have portrayed all of our feelings about her any better.
- Pa & Nanny



GRAMMY AND KEIRA

Keira is my "Sunshine" and my hero. She inspires me to love and understand more deeply. I am filled with a special kind of joy when we are together.



RISSIE WITH GRANDMA AND FAMILY

Rissie is my bright light. She is funny, smart and loving. The fact that she has Retts hasn't stopped her from living her life. I am truly blessed to be given the opportunity to share this life with her. Could a grandma ask for more?
- Maureen Tito



NANA AND JAYLA

Jayla is such a joy, an encouragement, so courageous, she can light up a room with her smile, flirt with her eyes, get in your face - grit those teeth to let you know that she is mad, try to get out of doing something with a kiss on the cheek, and make a whole room giggle with her laugh.



SIBLINGS

Having a sibling with Rett syndrome can pose its challenges at times but the love, compassion and strength that become apparent overtime is incredibly touching and quite remarkable.

Search for "IRSF Siblings Network" on Facebook and meet other siblings!

IRSF applauds all siblings everywhere!



Reagan and Lauren



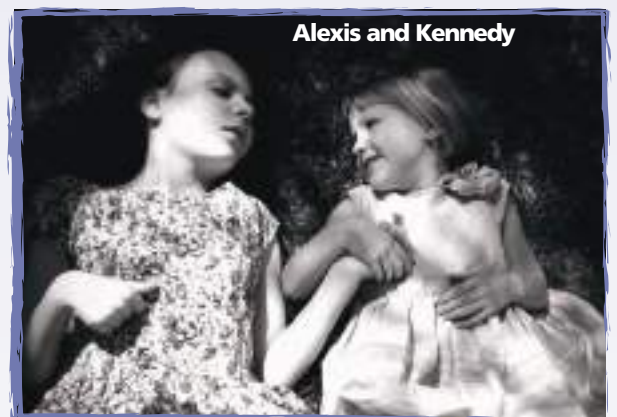
Olivia and Brianna



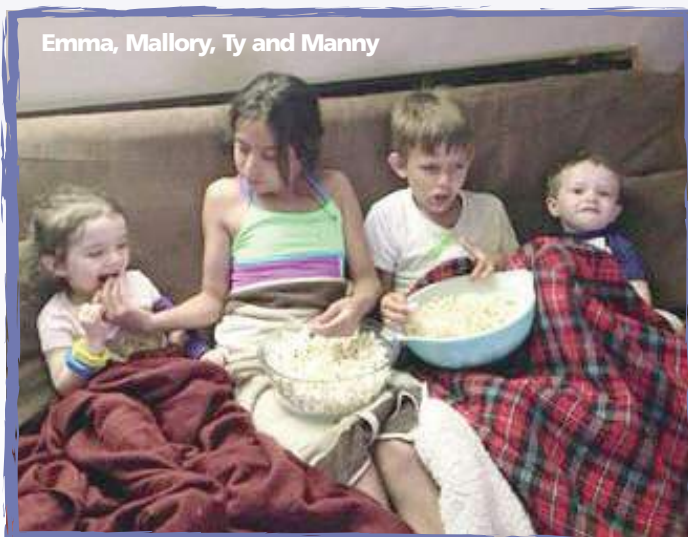
Emma and Hannah



Betsy, Maddie, Paige and Ellen



Alexis and Kennedy



Emma, Mallory, Ty and Manny



Izzy and Sera

Get Connected TODAY!

IRSF has grown to over 13,000 fans on Facebook so far this year! Get connected! Join our Rett syndrome community on our Facebook Fan Page. YOU can help us unite even more families on facebook! Direct all friends, family and NEW FAMILIES to the IRSF Facebook Fan Page to get connected!

Type "IRSF - International Rett Syndrome Foundation" in the search box and "like" the page. Use the wall to ask questions, share stories, comment on posts and to get connected with other families just like you!



Also, like your IRSF State Facebook Group Page to connect with families near you and stay up to date on local news, happenings and events! Families near YOU are connecting right now on Facebook! Just search "State" – IRSF Rett Syndrome Families to find your state's page and request to join! The majority of these pages are closed groups to protect the privacy of the sensitive subject matters that pertain to Rett syndrome. Contact lgeglein@rettsyndrome.org for questions or to start a state page if your state does not currently have one! Thank you in advance for taking the lead in uniting your community.

MORE WAYS TO CONNECT

- Subscribe to receive the IRSFlash for monthly updates sent right to your inbox to keep up on what's going on in the world of Rett syndrome.
- Follow IRSF on Twitter for breaking news and join the conversation! Get Rett syndrome trending by including "#IRSF #rettsyndrome" in all of your posts and tweets.
- Keep current by contacting IRSF to ensure that we have your correct contact information. If you need to update your address, email or phone number, please email lgeglein@rettsyndrome.org or call 1-800-818-7388.



SEARCH IN THE SPIRIT OF GIVING



Have you downloaded the International Rett Syndrome Foundation TOOLBAR, a new way to support our organization? With The IRSF toolbar, each time you visit and purchase items from over 2,100 popular sites such as Amazon, eBay, Expedia and Walmart.com, a donation will be made to IRSF.

DOWNLOAD NOW!

Visit <http://rettsyndrome.donation-tools.org/rettsyndrome/DirectDownload.aspx> to download the IRSF TOOLBAR and continue searching and shopping as usual.

Please open in **Firefox** (click allow), **Internet Explorer** (click run) or **Chrome** (click continue) to accept.



WEAR THE AWARENESS!

Check out IRSF's merchandise and spread awareness everywhere you go! From bracelets to t-shirts, hats to car magnets, IRSF has just what you need to help spread awareness. These items also serve as great gift ideas! Just visit the IRSF store and wear the awareness!

Featured Angel

Be a part of IRSF's monthly Featured Angel. IRSF spotlights one of our many inspiring Rett angels in the monthly IRSFlash! The Featured Angel's intent is to give our Rett community an introduction to a few of the Rett angels for whom we are fighting to turn Research into Reality.

LEAH LAYTON

Hi! My name is Leah Layton. I was born February 22, 2007 in Palo Alto, California. My first year was a dream. I ate and slept so well and learned to do lots of things on my own, including saying lots of words, phrases, and animal sounds. I fed myself and loved to read books. It took me a little longer to crawl, cruise and walk, but by 26 months I was running. Around the same time I started walking, my mom and dad noticed a few words disappearing. I was enrolled in an Early Start program and Teacher Jan suggested I get tested for Rett syndrome. So I did, and our lives were never the same.

Over the next few months, Rett syndrome took my words and hand use, but it didn't take my spirit. Mom says my smiles and giggles can still melt anyone and dad says my bright blue eyes twinkle on the darkest of days.

When I was first diagnosed, we were lucky enough to live close to Katie's Clinic in Oakland, CA where we received fantastic care and met some wonderful people. When I was four, dad decided to go back to school to get his MBA, so we moved across the country (I was in the car for SIX days!) to New



Hampshire where he attended the Tuck School of Business at Dartmouth College. On our way, we were able to meet even more wonderful Rett families. After dad graduated, we all moved again. This time, we just moved halfway across the country where we settled in the Midwest (Quad Cities, IA/IL), where we've met even more friends!!

My little sister, Kate, joined the family about a year and a half ago. She used to be kind of boring and just cried a lot, but she is getting to be more fun every day. And she gives me the best kisses.

I love spaghetti and swimming, my baby sister and sunshine, playing in the snow and swinging. Kung Fu Panda is probably the best movie ever made and, with my long blonde hair, I look a lot like Rapunzel. I love Taylor Swift and have been a big fan of hers since I was two. We have dance parties daily at our house so I have some pretty sweet moves. I still love to read and some current favorites are The Magic Treehouse Series, books about horses, and fairy tales.

Read more about Leah and all of the IRSF Featured Angels at rettsyndrome.org, News & Media-IRSF Features.

CONTACT

lgglein@rettsyndrome.org
to add your angel's story.



Angels from L to R:

MELIA, GABRIELLA, EVE, KAILA, CRISTIA ISABEL, ALYSSA, AVERY, ISABELLA, ALICIA



VERY SPECIAL EVENTS HOSTED BY VERY SPECIAL PEOPLE



Pictured from the top: St. Louis, Cincinnati Tristate, and Woodlands Strollathons



2013 STROLLATHON SEASON

The 2013 Strollathon program is now closing in on \$1 million raised. Strollathons started out strong in the spring with the first three Strolls in Chicago, Michigan and St. Louis raising over \$186,000 – an average of over \$60,000 per Strollathon! Thank you Stroll chairs Shelby Swann, Lisa Fenberg, Joyce Opinsky and Amy Kreher for a great kick off to the 2013 Strolls. After a 3 month hiatus, the fall Strollathon season really began to heat up with 10 Strolls in September, 9 in October and finishing out the year with Strollathons in the Woodlands, TX and Las Vegas, NV.

The Cincinnati Strollathon celebrated its 10th Anniversary with a cake and cupcakes for all. Over the years there have been a number of event chairs and in 2013 Jennifer Braner, Tracy Bergman, Lisa Byrne, Paula Bell, Anne Rich, Jodi Rodrigues, Kathy Ballmeyer joined forces to raise over \$135,000 and bring the total raised since 2004 to just over \$2 million. Congratulations to these very special people.

Petrina Pate has been chairing the Roanoke, VA Strollathon for 7 years now and due to all of her hard work, including her Meet and Greet Kick-off, she raised over \$42,000 – her biggest year ever – way to go Petrina!

Choo Smith, our honorary National Chair of the Strolls appeared at both the Baltimore and National Capital area Strollathons which together raised almost \$60,000. Thank you Carol Hulett, Monique Ray, Laura Gomez, Lisa Ruane and of course, Choo Smith.

Evie Swan took on the huge task of chairing Strollathons in Reno and Las Vegas, NV as well as in Augusta, ME. The combined events raised over \$66,000. Thanks so much for all you do and your endless energy, Evie.

The Florida Strollathons to date have raised just shy of \$100,000 this year due to the hard work of co-chairs Henry Perez, Elizabeth Allocco, Charity Proffitt, Nikki Johnson and Barry Rinehart. Since 2010, the Florida Strollathons have raised over \$380,000! Congratulations to all!

Jennifer and Justin Endres hosted the 8th Annual Cape Cod Strollathon in September and over the years has raised over \$841,000 for Rett syndrome. I'm sure when they host the 10th annual Cape Cod Strollathon, they will reach the \$1 million mark. Go Jennifer and Justin!

The Nebraska Strollathon hosted by Jacy Horst was held on September 21st in Grand Island. This was the second Stroll that Jacy had chaired and she has already raised over \$135,000 in just two years. Great job, Jacy.

Shannon Stoiber, along with the help of her mom, Deanna Pence, has chaired the Olathe, KS Strollathon for the past four years and has raised over \$76,000 – thanks for all you do Shannon.

This is the second year that Patty Mevis has chaired a Strollathon in Green Bay, Wisconsin – in 2012 \$26,344 was raised and so far in 2013 the stroll has raised \$26,322 – just \$22 shy of last year – can someone help push her over the top? Way to go Patty!

Ashley Alexander and co-chair Gail O'Quinn hosted the 3rd Annual Georgia Strollathon in Macon in October and raised almost \$25,000. They also hosted a wonderful kick-off swim party for participating families and made and distributed many wonderful and creative personalized items. You are so appreciated!

The Portland Strollathon is in its 8th year - Joey Razzano has chaired each one and has raised over \$200,000 since 2006. Joey had to rush back from the family's Make a Wish cruise to Bermuda to put the finishing touches on this year's stroll. Don't hesitate to ask her about the cruise – it was an amazing experience for Jade and all of the Razzanos. Thanks Joey!

The Northern California Strollathon chaired by Erica Robertson is in its 5th year and has raised over \$232,000 in that time. For the past couple of years, the Strollathon was done in conjunction with the Oakland Natural History Study which has given many families on the west coast the opportunity to experience a Strollathon. Thanks for all you do Erica!

Leslie Greenfield, president of the New Jersey Rett Syndrome Association, hosts an annual gala and golf tournament in New Jersey and for the past 5 years in her spare time, she has chaired the New Jersey Strollathon! This year the stroll raised over \$32,000 – one of her best years ever. We thank you Leslie!

Southern California families have been supporting their local Strollathon chaired by Sherri Brady, since 2010. To date, the SoCal Stroll has raised over \$125,000 – thanks for all you do for the SoCal families, Sherri.

Texas held Strollathons in San Antonio, Grapevine and the Woodlands in 2013 hosted by Kenna Seiler, Amber Weigl Vanessa Peace and Carrie Weaver. So far the Strolls have raised almost \$60,000 as money continues to come in. What a great team!

The Grand Rapids, MI stroll was held in September and chaired by Nicole Pintoski and Nicole Mast. This was their second year for this Michigan Stroll which has raised over \$27,000. Keep up the good work ladies.

Thank you to Mary Kallstrom who has been hosting the Minnesota Stroll for the last four years. We appreciate your tireless efforts and enthusiasm. Her event has raised over \$63,000 since the inception.

We'd like to give a huge shout out to Randstad and Jackie Piscatelli. Jackie, the liaison between Randstad and IRSF, attended and was a very busy volunteer at Strollathons in Chicago, Georgia, Florida and Cape Cod. Randstad employees took up our cause and raised money for all of these strolls as well as events in Baltimore, Cincinnati, Northern and Southern California and Texas.



Pictured from the left to right: Chicago, Roanoke, National Capital, Baltimore and Portland Strollathons



Fore!

Golf is a passion for many people and also the preferred way for a number of our families to raise funds for Rett syndrome while enjoying their favorite pastime.



Samantha Corpus Golf Tournament

The golf season opened in May with two hugely successful tournaments hosted by IRSF Board members. Mike Joyce hosted the 11th annual event at the Blackrock Country Club in Hingham, MA and John Corpus hosted the 6th Annual Samantha Corpus Golf Tournament at the Blackhawk Country Club in Danville, CA. Thanks Mike and John for all your hard work on the course and on the board.

The season continued with Jim Bathie's 3rd Annual Cure for Kiera Golf Tournament which celebrated its biggest year ever. Way to go Jim – we know how excited you were to have such a great success.

Leslie Greenfield, who seems to never rest, held the 4th Annual New Jersey Golf Tournament at the Battleground County Club in Manalapan, NJ. Thanks for ALL you do!

The golf season closed out with the 2nd Annual Mark Charamella Memorial Golf Tournament hosted by Debbie McLaughlin in memory of her father and in honor of her sister Deirdre who has Rett syndrome. What a wonderful way to honor your father's memory and to help IRSF continue its mission.

Pat and George Fay have been hosting their golf tournament in honor of their daughter Meghan for 17 years now. This year's event was at the Portland Golf Course in Portland, CT. Thanks to both you for all of your hard work.



Charlotte Troy Golf to Cure Rett Syndrome

The 5th Annual Charlotte Troy Golf to Cure Rett Syndrome was held in September at the Southerns Marsh Golf Club in Plymouth, MA hosted by Rick Tory. Keep up the good work Rick – we really appreciate your efforts.

There were several other golf events held throughout the year that will be mentioned in our grassroots event coverage.

GALA EVENTS

The foundation sponsored two large gala events this year. For over 15 years, the foundation has hosted an evening event in New York City. There have been many chairs of this event – for the past 10 years it was hosted by Maura and Steve Gallucci - and this year Sean and Lisa Lebson along with Robin and Eric Diamond and Alissa and Barry Schurr took the event in a new direction and to a new venue. A great time was had by all in attendance at the Reach for the Stars event and they raised over \$265,000 for Rett syndrome. Thanks to all your hard work to continue one of our longest running events.

Board Chairman, Rajat Shah, and his wife, Jenny, along with FSB Chair Suzie Solender and her husband Peter, hosted the 3rd Annual Hustle for a Cure at the Harbour Club in the First Niagara Center in Buffalo, NY. Guests enjoyed an evening of great food and entertainment and a live and silent auction while raising over \$200,000. Thanks for all you do!



2013 Reach for the Stars



Left to right: Birdies for Bennett, Course for a Cure, Heather's 40th Birthday Fundraiser, HER Night Fiesta, Laughs for Livi Bets for Bella

GRASSROOTS EVENT LIST

1st National Jeans Friday	First National Bank in Fairfield	HER Night Fiesta	Dora and Daniel Castner
2nd Annual Paddle For Sorel	Megan and Chris Kolendrianos	Her Night Happy Hour	Robin and Eric Diamond
31 Fundraiser	Emily and Alfonso Estades	Heroes of the Silent Angels Tournaments	Catherine and Solomon Galyon
A Lil Bit of Fun - 7 Iron Challenge to Cure RTT	Gina and Ryan Mueller	Jean day	St. Patrick Parish
AEO Backyard BBQ		Jeans Day	St. Anthony Grade School
IHO Miya Zerial	Kimberly K. and Charles LaClear	Katelyn's Cookin' Butts for Rett Syndrome	Mrs. Kristi Byers
Annual Summer Solstice Party	Marianne and Mark Kozub	Knight Garage Sale	Ms. Sandy Johnson
Baran's Kenpo Karate	Baran's Kenpo Karate, LLC	Laughs for Livi, Bets for Bella	Jill Twigg and Jeff and Holly Rutko
Basket Raffle	Robert W. Baird & Company	Lemonade STAND for Rett	Ariane and Christian McMahan and Family
Bicycle Tour de BrewSA	Mr. and Mrs. Robert Stilwell	Live Rett Free	Karen Herald
Birdies for Bennett	Dr. Margaret Dahms	Meals for Maddie	Mrs. Jessica McKnight
Black & White Affair	Kay and Michael Borchardt	Men Are Pigs	Carol and Andrew Hollowell
Blue Jeans 4 a Cause	Mrs. Susan Lee	Morgan Mize Fundraiser	Francis and Jeff Mize
Blue Olive Fundraiser	Mrs. Kaycie LaGrone	Nets for Rett	Ms. Dawn Moses
Bruins Alumni Game	Tom Guidi	Oakley's Day of Fun	Mrs. Shannon Madden
Carpenter Calendar	Trisha and Steven Carpenter	Origami Owl	Laurie and Eric Wells
Ceirney's Ride	Tammy and Chuck Wilson	P.U.S.H	North Brunswick Township
Charity Rope Halter	PNW Select	Preceptor Beta Iota of Beta Sigma Phi	Anita and Brent Poulton
Chicago Run	Diane and Mick Ross	Quaker Steak & Lube Fundraiser	Dr. Curt Heide
Choose Your Charity	Foxborough Public Schools	R.O.A.R - Rock out Against Rett	Sally and Josh Teague
Course for a Cure	Jennifer and Brian Hockings	Race for Grace	Tara and David Reddington
Dance for a Smile	Lucrecia and Andrew Hoepfner	Race for Mace	Mrs. Michelle Walker
Donate to Dress in Denim	Childrens Medical Associates of N. Virginia	Rett Rendezvous	Misty Startup
Dress Down Friday	Western Regional Jail	Rett Ride	Angie and Brad Weaver
Dress in Denim Day	West Essex Middle School	Small Change for Change	Mr. Matthew Nelson
Emery's Birthday	Carrie and Barrett Fowler	SpaTacular Party	Raquel and Antonio Priolo
Emma Empowered	Mrs. Karen Irizarry	Spike it for Rett	Guess, Cooper and Brunner Families
Environmental Ladies Who Lunch	Amber and Dan Weigl	Stoller County Fair	Nancy and Herbert Stoller
Erica Foreman	Miss Erica Foreman	Strollathon	Mrs. Yaritza De Leon Torres
Erika's Walk	Mary Lou and Gary Miller	T Shirt Sales	Heather and Nick Kauffman
Fight for Alex	Michelle and Joshua Poulsen	Token Auction	Darcey and Doug Garno
Find a Cure for Rett Syndrome Fundraiser	Ms. Jessica Seiber	Valentine's Appeal	Charleen and Lawrence Ryan
Galatas Elem. Fundraiser	Galatas Elementary School	Walk and Roll for Rett	Susan and Ronnie Brackett
Genetic Research Project	Chelmsford High School	Walk-a-Mile-in-Havana's-Shoes	Mrs. Chantele Leatherwood
Get Rad for Rett	Mr. Alexander Dones	We will hear her voice!	Mrs. Ann Marie Brady
Golf Scramble	Ms. Bridgett Lindsey	Wolf Walk	Mrs. Rebecca Brasher
Green Tree Festival	Pretzel Boys LLC	WVU Hospital Employee's	Ms. Donna Corley
H.O.R.S.E Benefit Horse Show	Syndi and Scott Blunk		
Haunted House Bake Sales	Mrs. Jennifer Castro		
Heather Cooper's 40th	Heather and Jason Cooper		

A DECADE OF HOPE

Many years back, a small group of co-workers in the San Diego workers' compensation community pooled their friends, families and community resources in the hope they could shine some light and awareness on a devastating disorder that affects the lives of precious little girls. At that time we had a co-worker who had 2 children with Rett syndrome and we were so overwhelmed with a need to help in any way possible. After much discussion on what type of event could raise the most money, we committed to an idea and started down a road we had no idea would be such a wonderful adventure. On October 8, 2004, the inaugural golf tournament took place and a new tradition in our small San Diego workers' compensation community was born.

This last July we celebrated Operation Hope, our 10th Year Anniversary and had a fantastic turn out once again. Over the past 10 years, we have raised over \$550,000, which, through IRSF, has been used to explore new treatments and hopes for a cure. The families, volunteers, sponsors and supporters have truly made each tournament more memorable and successful and are the sole reason we have been able to continue this very awesome tradition. Over 400 participants return year-after-year to be a



Decade of Hope Volunteers

part of this community experience and enjoy the opportunity to meet the families and bring them some hope for a cure. The same Rett families come year after year and are like an extended family to all of us. Watching these girls grow each year drives us to continue the efforts to raise funds so these little angels may have a brighter future. We have all been blessed by this opportunity to raise money for Rett and hope to continue the tradition for years to come.

If you have any interest in participating as a volunteer, sponsor or golfer, please visit our website at www.greenenvyink.org.

--Tournament Directors, Deirdre Gonzalez & Sara Mulick

A message from A PASSIONATE GRANDMOTHER ABOUT AN AMAZING GROUP OF LIFE-LONG FRIENDS

When my five-year-old granddaughter was diagnosed with Rett Syndrome three years ago, our family went from teaching this beautiful child about the world to teaching the world about her. We hardly had time to dwell on the sad and frustrating feelings that surround this condition; our friends and my daughter's friends jumped in immediately to help. They offered prayers, listening ears, strong arms and open hearts for emotional support, helped with fundraisers, educated themselves about Rett syndrome and donated to the Michigan Strollathon.



Jolan's lifelong friends and Rett syndrome supporters

Among these serious Sydney supporters is a group of wonderful women who have been my friends for over 50 years. Pals from grade school, church and scouts, we attended the same high school, where we began calling ourselves the Vegas. We have grown closer with age and life experiences, and many travel from all over the U.S. to attend our three-day gathering every summer. From college days, career and child-raising days -- when we only had time for quick, occasional lunches -- to spending long weekends at my lakeside cottage, our collective relationship has blossomed into an anchored connection; a safe and happy place where we share, support, be silly, laugh and love . . . no matter what! And so my dear friends were more than eager to smile for this picture for Sydney Jolan (see how she shares my name!) and support the International Rett Syndrome Foundation.

– Jolan Webster

\$100,000 Gift from RSAM Funds a Project to Model Rett Syndrome in Stem Cells



Rett Syndrome Association of Massachusetts recently made a \$100,000 gift to IRSF designated to fund a research project called Modeling Rett Syndrome Using TALEN Technology in Human Pluripotent Stem Cells.

The project is led by Dr. Yun Li at the Whitehead Institute at MIT and aims to establish new models of Rett syndrome using genetically engineered human pluripotent stem cells to serve as an unlimited source of disease-specific human cells. The cells will be used to study the molecular, cellular, biochemical, and electrophysiological aspects of Rett syndrome. Moreover, the cells can be tested for how they react to existing drugs at a very fast pace. In fact, in a recent publication by Dr. Li, this research described these new cellular tools and demonstrated their use in testing compounds (such as IGF-1 and BDNF). The paper points out that IGF-1 therapy is one that has great potential and illustrates the wisdom behind our support of the current clinical trials. IRSF is excited about this project as yet another avenue that may help to accelerate the process of finding effective relief for children affected by Rett syndrome and their families.

Rett Syndrome Association of Massachusetts is a longtime partner of IRSF, yet this year's commitment brings the IRSF-RSAM partnership to yet another level. We believe that together, we can reach our common goal of finding therapeutic strategies to prevent, delay, and reverse Rett progression while we develop treatments and a cure for Rett syndrome faster.

To learn more about RSAM, please visit www.rettsyndromemass.org.

CLAIRE'S CRUSADE PARTNERS WITH IRSF TO FUND CUTTING EDGE RESEARCH

Claire's Crusade, a 501(c)(3) non-profit organization based in Cleveland, OH, has grown quickly since its founding in 2012. Just recently, they proudly contributed \$75,000 to the fight against Rett Syndrome, with a \$25,000 gift designated to support IRSF's cutting edge translational research program.

"This gift is sure to be the first of many and we're excited that it marks the creation of an expansive partnership between organizations driven to improve the quality of life of those battling Rett Syndrome. This is really just the beginning for us at Claire's Crusade – it's really just scratching the surface in terms of the impact we plan to make as we continue to grow."

- Sean M. Reilly: Claire's Crusade Co-Founder, President

IRSF welcomes this partnership as a great opportunity for collaboration on our quest to develop a cure for Rett Syndrome. By working together, we can accomplish more, faster.

Claire's Crusade is dedicated to raising funds in support of cutting edge research efforts to develop treatment and a cure for Rett Syndrome and is equally committed to increasing awareness and providing resources for families faced with the uncertainty this affliction can bring.

Visit ClairesCrusade.org or 'Like' them at [Facebook.com/ClairesCrusade](https://www.facebook.com/ClairesCrusade) to learn more.



BREAKING TRADITION FOR IRSF

The walls of Judge Bean's Restaurant and Cantina are covered with money. Keeping with tradition that dates back to the wild west, customers write messages on a dollar bills and attach them to the wall. Legend tells us that in the 1880's, outlaws at the first Judge Roy Bean's Saloon in Langtry, Texas used this "money posting" as a way to communicate with a friend who might be passing through the town. Judge Bean, sometimes known as "the hanging judge", held court in his saloon in this little Texas town. He called himself the "Law West of the Pecos". Today, the owners of Judge Bean's in Keller, Texas, Al Said and Bill Mena, continue the money posting tradition. But unlike the first Judge Bean, they don't dish out justice, just good food and a friendly atmosphere for their customers. Since the Keller restaurant opened in 1992, customers have enjoyed writing messages on their dollars to hang on the wall, so the money has continued to accumulate. Said and Mena have only removed money from the walls twice since 1992- once to assist the victims of 9/11, and once to contribute to the International Rett Syndrome Foundation. Their gift to IRSF makes them a Silver Level Sponsor of the Grapevine, Texas Strollathon, which was held on October 27, 2013. Frequent customers of Judge Bean's Restaurant and Cantina, Keith, Lynne, and Allison Buchwald (RTT), first introduced Said and Mena to IRSF. Both Said and Mena are pleased that their donation will be used for research to find treatments and a cure for Rett syndrome. Allison Buchwald is pictured with Al Said, the wall of money, and the certificate of appreciation from IRSF.



Allison Buchwald and Al Said