Rett Gazette
SPRING • 2013
INTERNATIONAL RETT SYNDROME FOUNDATION

IRSF
INTERNATIONAL RETT SYNDROME FOUNDATION

Message from IRSF Executive Director

Stephen E. Bajardi

Hello to our Community,

IRSF and the Rett syndrome community finished 2012 with strong momentum. Our national board of dedicated volunteers, most of them Rett parents, approved a second round of research grants for an annual total of $3.7 million in direct research support. In 2012, IRSF hosted a successful World Congress, bringing together families, Rett syndrome organizations, and scientists from around the world. 2012 also saw the continuing support for the IGF-1 trial at Children's Hospital Boston, new support for the NNZ-2566 trial at Baylor College of Medicine and a preclinical project at Nemours/Alfred I. DuPont Hospital for Children, as well as support for basic science and other treatment related research.

Looking ahead to 2013, it will be a year of change and growth for IRSF. The national board is conducting an extensive search for new board members; people who are touched by Rett syndrome, have a strong commitment to fundraising and to being stewards of an organization searching for more treatments to reduce the effects of symptoms on children and adults with Rett syndrome. We expect challenges ahead with added demands on IRSF as federal sources for research funds stop growing and investigators look to us to fund new projects. We will likely see an increased interest in our research program of grants, fellowships, and contracts.

We are optimistic and excited about increased funding of translational science in 2013 with IRSF directing more scientific resources towards finding new treatments. There is enormous scientific interest in Rett syndrome, especially in the area of treatment related research; something we've encouraged for a long time. We will do everything within our power to fuel that momentum because it presents an opportunity for more attempts at identifying compounds as potential medicines for Rett syndrome.

One thing is certain, IRSF is passionately moving ahead with the priority of improving the lives of children with Rett syndrome as quickly as possible. Our approach to Rett syndrome is different. Driven by our mission to provide family support and fund research, we look at the totality of the child and family touched by Rett syndrome. Our staff includes parents of children with Rett syndrome and that allows us to better understand and connect with the needs of the community. By directing scientific resources to areas of research for the greatest impact on their lives today, we are meeting a unique niche in Rett syndrome research. Our family support program covers the complexity of Rett syndrome and offers services tailored to the Rett syndrome community. We host special interest groups like Dads, Grandparents, Siblings, Spanish Speaking Families, Educators, and others. We provide information, support and networking through our social media program, RettNet, and the annual family conference. Our volunteers at the state level, the Regional Representatives, continue to be the backbone of providing the peer to peer support necessary to help families acquire the best resources available for their children. The Family Advisory Board, led by volunteers with firsthand experience with Rett syndrome, continues to take on major advocacy and public awareness projects such as October Rett Syndrome Awareness Month, Rare Disease Day, and the lobbying of TV and radio stations to play the Clint Black PSA.

> *Driven by our mission to provide family support and fund research, we look at the totality of the child and family touched by Rett syndrome.*

It is because of all of you, the families that donate to IRSF, raise funds, advocate, and educate, that IRSF can purse this path. Our Strollathon program is growing, led by volunteer chairs on the ground, raising more money and getting more new families involved than ever before. The financial impact of Strollathons is huge, as is their ability to organize and unite the community in its common interests and goals. IRSF is expanding its capacity to support our volunteers, the remarkable people who host golf events and galas and those who train and run for new pledges in the Rett Racers program. Community family fundraisers and donors who make many personal sacrifices continue their financial

continued on page 2

The bond between sisters needs no words.

leadership

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.

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.

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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.

Family Advisory Board Message and Update

Dear IRSF Families,

I am delighted to be chairing the 2013 Family Advisory Board (FAB) and pleased to write that we started the year off with vigor in January. Your FAB members are Joy Rosen (Chair), Connie Lindberg, Clifford Fry, Shawn Mansfield, Vicki Kirihara, Marie McTernan, Donna Wright, Suzie Solender, Lynn Peel, Deberah Patterson, Daniel Warner, and Maureen Lee. We have some new members joining us and look forward to introducing you to them.

Our 2013 focus of interaction with the IRSF Staff and Board of Directors is to bring you exciting new fundraising and awareness events. Our members are brimming with "out-of-the-box" energy to bring us beyond our financial goals for research and to spread awareness of Rett syndrome across the country.

The **Advocacy and Awareness Committee** continues efforts to obtain Proclamations for EVERY State and expanding ideas for utilizing the Proclamations to increase fundraising. They are also working to strengthen enrollment in the Natural History Study and will be developing Community Awareness programs that YOU can use to educate your local teachers, doctors, therapists and general public about Rett syndrome.

The **Information and Communication Committee** will focus on updating Rett syndrome informational materials and develop strategic ways to keep moving IRSF forward within the world of social media.

The **Family Support Services Committee** is already hard at work to bring you an exciting and worthwhile Family Conference in Utah with new exhibitors and enjoyable activities. Also, watch for exciting news about expanding our Rett Community Network Groups.

And of course our **Fund Development Committee** is hard at work looking into new opportunities to bring creative, fun ideas to meet our much needed fundraising goals for critical research.

We are here to support, encourage and engage each of you in our efforts. We need volunteers throughout the year to help on small and large projects. Please email me joy.rosen@hotmail.com and let us know where and how you'd like to help.

Sincerely,
Joy Rosen

Message from IRSF Executive Director continued

support and advocacy to power the momentum of research and increase family support.

IRSF benefits greatly from the support of regional Rett syndrome associations from New Jersey, Minnesota, Colorado, the Pacific Northwest, Illinois, Massachusetts and the Southeast. These wonderful allied organizations are dealing with local needs supporting clinics, families, health care providers, and national programs such as the annual family conference and the Natural History Study, a program vital to translational research.

IRSF also acknowledges the support from numerous private family foundations, the 12 specialty Rett syndrome clinics in the U.S. that are closely aligned with the IRSF mission to keep our children healthy and investing in clinical research; all of the international Rett syndrome associations that we work in concert with daily; as well as our friends with MECP2 duplications, MECP2 mutations, and related disorders such as CDKL5 and FOXG1. Working closely with other associations fosters synergy in the research field, sharpens their focus, and improves the lives of families everywhere. It truly takes a village to succeed.

Your national board is committed to the mission of bringing relief to children living with Rett syndrome. Our volunteer leaders, our staff, and I are committed to each and every one of you; your passion is contagious and the heroism of the community is inspiring. We will work together to make 2013 the best year ever. You all are entitled to nothing less.

Warm regards,

Stephen E. Bajardi

Message from IRSF's Chief Science Officer

Dr. Steven G. Kaminsky

Greetings:

Wow it has been over a year since joining IRSF, and it has been a fast and furious ride. The learning curve has been steep but very rewarding for me. I have made great new friends, met wonderful families and fell in love with a group of angels!

Looking back on this first year there have been many highlights: The 7th World Congress in New Orleans brought families, clinicians and scientists together to review successes, learn from each other, and plan for the future. The vision by all was inspiring and reminded me of an Abraham Lincoln quote, "Determine that the thing can and shall be done, and then we shall find the way".

Today we all work to chase Rett down. We look for better ways to treat the symptoms, we explore for methodologies to change the pathology, and ultimately we continue to search for ways to reverse the disorder and free our children from the constraints that MeCP2 has placed on Rett patients. As I write this note, patients are being enrolled into two studies that are designed to circumvent the pathology associated with Rett syndrome. The Boston Children's group under Dr. Walter Kaufmann and the Baylor College of Medicine group under Dr. Daniel Glaze bring us hope that these two clinical trials will provide an opportunity to help our Rett families with new and unique therapies.

At the same time we continue to invest in other novel discovery programs and therapeutic programs that push the horizon in new directions. Drs. Steven Gray and Kevin Foust are expanding the possibilities of future gene therapies; Drs. Dag Yasui, Jay Shapiro, and Kevin Jones are identifying new therapeutic possibilities to modify the biology associated with Rett syndrome; Drs. Beth Stevens and Ali Khoshnan explore the role of the immune system and microglial cells as potential players in formulating new therapeutic strategies for Rett; and finally Drs. Alysson Muotri and David Lieberman are developing methodologies to screen larger libraries of drugs for therapeutic potential in Rett syndrome.

All of this paints a wonderful horizon, and it reminds me of a second quote from Abraham Lincoln, "Commitment is what transforms a promise into reality". We at IRSF have always promised to do all we can for the families and girls diagnosed with Rett syndrome. We are committed to this promise and will continue to push in any way we can to bring a new reality to treatments, caring and providing for our Rett families.

Sincerely,

Steve Kaminsky, Ph.D.

RECENT UPDATES ON CURRENT RETT SYNDROME CLINICAL TRIALS

Phase 2 Clinical Trial of IGF-1 is approved to move forward with patient recruitment

Boston Children's Hospital has received approval to move forward with Phase 2 of the clinical trial entitled "Pharmacological Treatment of Rett Syndrome by Stimulation of Synaptic Maturation with IGF-1" which allows them to begin actively recruiting subjects. IRSF congratulates the team for their perseverance and commitment to improving the lives of those with Rett syndrome.

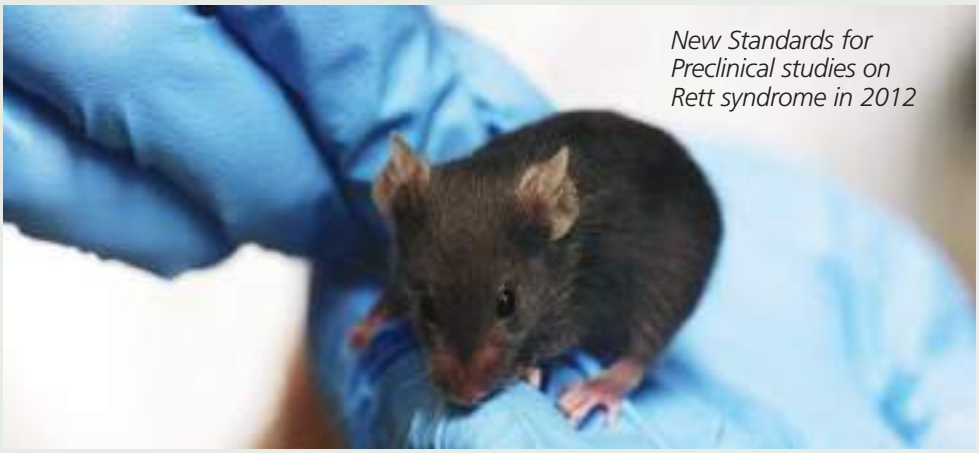
Interested families may begin the pre-screening process to determine the eligibility of your child by contacting the Rett Syndrome Research Program at BCH by email (rettresearch@childrens.harvard.edu) or phone (617-355-5230).

FDA Approves the IND for the Phase 2 Clinical Trial study of NNZ-2566 in Rett Syndrome

The US Food and Drug Administration (FDA) has approved the Investigational New Drug (IND) application in January 2013, allowing Neuren and Baylor College of Medicine to proceed with the Phase 2 clinical trial to assess the safety and efficacy of NNZ-2566 in adolescents and adults with Rett syndrome. This is the first clinical study in older patients diagnosed with Rett syndrome. Baylor College of Medicine has begun screening patients based on their inclusion/exclusion criteria, and enrollment.

Interested families may contact the project manager Aryn Knight at the Baylor College of Medicine by email (apolk@bcm.edu).

2012 Research Highlights



New Standards for Preclinical studies on Rett syndrome in 2012

MeCP2-deficient neurons do not respond to respiratory distress nor increase BDNF.

IRSF funded investigator John Bissonnette, MD and Agnieszka Balkowiec, MD, PhD of the Oregon Health & Science University have published new findings on the regulation of brain-derived neurotrophic factor (BDNF) by MeCP2 in the March 2012 issue of Neuroscience. Dr. Bissonnette and his colleagues found that the mutant neurons in a RTT mouse model failed to increase BDNF in a paradigm that markedly increased the neurotrophin in normal mice. In their experimental study, they exposed the animals to an environment of low oxygen to challenge the respiratory system. They found that normal neurons in the areas of the brain that respond to respiratory signals will dramatically increase BDNF upon low oxygen exposure, whereas BDNF protein expression in mutant neurons that lack MeCP2 remains the same.

Respiratory issues are just one symptom that those diagnosed with RTT may contend with. In light of these new findings, low oxygen levels during apneas or other breathing issues may fail to induce BDNF in the respiratory regions of the brain, which then may intensify respiratory dysfunction. Therefore, increasing BDNF levels or its function is a highly studied area to find a potential therapeutic for RTT.

Eye-Tracking Technology shows Social Preferences in Rett Syndrome

IRSF funded investigator Dr. Aleksandra Djukic and Dr. Maria Valicenti McDermott at the Rett Syndrome Center at Montefiore Medical Center have published their results in the April 2012 issues of Pediatric Neurology on their exciting study of girls with RTT and whether their strong eye gaze can indicate their preferences and be used as a mode of non-verbal communication. This pilot study investigated 49 girls genetically diagnosed with Rett syndrome between the ages of 1.5-25 years and examined their pattern of visual fixation (eye gaze) using a Tobii eye-tracking device that displayed various pictures and monitored where on the picture the girls would look and fixate. This device also determined how long the girls looked at these pictures. The results of this study indicate that "girls with Rett syndrome exhibited a preference for socially weighted stimuli". The study participants would focus more on people in the pictures, rather than any other objects (e.g. a fountain or a statue within the same picture). A picture containing people would be looked at more times than an adjacent picture of a red dress. When a picture of a person's face is shown, the girls would focus much more on the eyes rather than the nose or mouth. A comparison with the control group of 33 typically developing girls showed that the females with Rett syndrome were more likely to look at the eyes of people in the pictures. This study also underscores a real difference in the visual attention of girls with Rett syndrome versus children with autism, who tend to avoid eyes and face. This suggests that girls with Rett syndrome can both process and prioritize – i.e. learn – information differently than children with autism.

MeCP2 is critical for maintaining mature neuronal networks and global brain anatomy during late stages of postnatal brain development

Dr. Nurit Ballas, who was awarded an IRSF Basic Research grant, published new findings in the July 2012 issue of the Journal of Neuroscience. The Zoghbi, Bird and Ballas groups have shown that loss of MeCP2 after birth, even at adult stage, gives rise to the same spectrum of severe symptoms as when MeCP2 was lost at birth. This suggests that the primary requirement for MeCP2 in the brain occurs at a stage when many neuronal connections are already formed. The extreme dependence of MeCP2 at this late stage of brain maturity was surprising, so Dr. Ballas and colleagues sought to perform an in depth analysis of the abnormalities to help explain why the brain required MeCP2 at this stage. Without MeCP2 at the adult stage, they found that brain growth was stunted where there was overall shrinkage of the brain, a fewer number of neurons, abnormal astrocytes, and a reduction in neuron cell dendritic branches and spines. Their work also revealed how critical MeCP2 is in the proper regulation of synaptic proteins, which is likely a direct requirement for the maintenance of mature neuronal networks in the adult brain.

The Origins of the Mutated *MECP2* gene: Sporadic mutation on the paternal X chromosome.

An interesting genetic case study of Rett syndrome in China that questioned the source of the mutant *MECP2* gene was published In the August 2012 issue of the European Journal of Medical Genetics. They found that mutations on the *MECP2* gene found in those diagnosed with Rett syndrome are more likely to come from the father's copy located on the X chromosome. This study analyzed 365 cases, where 315 had a *MECP2* mutation. Deeper analysis of parental origin in 139 cases found that 94.4% of these cases harbor mutations from paternal origin and the remaining 5.6% are of maternal origin. All of these mutations except for one case were sporadic mutations.

Ketamine Reverses Abnormal Brain Function in Rett Syndrome Mice

IRSF funded investigator Dr. David Katz and his colleagues at Case Western Reserve University published novel findings in the October 2012 issue of the Journal of Neuroscience that demonstrate the reversal of brain activity abnormalities and improvement in neurological function of RTT mouse models with ketamine treatment, an FDA-approved anesthesia drug. The authors sought out to create the first global map of neural activity in the RTT brain to understand the brain's circuitry dysfunctions in Rett syndrome. In order to do this, they compared brains from *Mecp2* mutant mice and normal mice by tracking markers of brain activity. These comparisons found that there are two regions in the brain where neural activity differed in the RTT brain. First, the forebrain, which controls nearly all processes in the Central Nervous System, was found to have less neural activity in the RTT brain compared to the normal brain. Second, the brainstem, which is responsible for basic vital life functions such as breathing, heartbeat, and blood pressure, was reported to have abnormally high neural activity in the RTT brain compared to the normal brain. By identifying that these brain structures have such different activity levels in the RTT brain, they were able to begin focusing on these areas to see if they could modify the abnormal activity.

Settings standards for research into Rett syndrome

In September of 2011, IRSF participated in a workshop focused on the state of the art in animal studies of Rett syndrome with the National Institute of Neurological Disorders and Stroke (NINDS) and other federal and private organizations interested in Rett syndrome research. A broad cross-section of basic scientists, clinicians, and representatives from the National Institutes of Health (NIH), the Food and Drug Administration (FDA), the pharmaceutical industry and private foundations attended the workshop in order to identify crucial knowledge gaps and to suggest scientific priorities and best practices for the use of animal models in preclinical evaluation of potential new RTT therapeutics. The outcomes of this workshop authored by several workshop participants and were published in the November 2012 issue of Disease Models & Mechanisms (DMM). The combination of an urgent need for effective treatments for RTT, coupled with the availability of good mouse models, is a driving force for studies that can identify and test new drugs.

5-hydroxymethylcytosine (5hmC): A Molecular Target of MeCP2

In the December 2012 issue of Cell, Dr. Nathaniel Heintz and his colleagues from the Rockefeller University published a new target for MeCP2 that may be important in its role in regulating genes. It has been well known that MeCP2 binds methylated DNA, hence its derived name "Methyl-CpG binding protein 2". The C stands for the DNA base Cytosine. With the relatively recent discovery of a related DNA based called 5-hydroxymethylcytosine (5hmC), researchers were eager to find out its role in genetics, especially because it is found highly enriched in the neuronal cell types. In this article, the authors identified that MeCP2 can bind DNA that contain either 5hmC or 5mC bases, but did not bind unmodified Cytosine bases. They further report that a specific Rett mutation was analyzed, R133C, a mutation that disrupts 5hmC binding to MeCP2, but only slightly effects 5mC binding. Further analysis on the transcriptional activity of MeCP2 needs to be performed to determine how these different bases affect MeCP2 function. However, the data presented in this paper support a model where MeCP2 binding to 5hmC-enriched DNA regions may lead to gene activation in neuronal cell types, while binding to 5mC-enriched DNA may not.

Updates on the Natural History Study

If you're interested in participating or learning more about this important study, please visit our updated website that now contains a new 6-min documentary directed by Joseph Mendoza that highlights successes from the IRSF-supported NIH Rett Syndrome Natural History Study. Check out the new document "Should I participate in the Natural History Study?" that can be downloaded from our website. The 2013-2014 site dates have been posted. We hope to see you there!

Check out the new Natural History Study video on

3rd European Rett Syndrome Conference Maastricht, "Research Update & Preventive Management" (ERSCM 2013)

This year's Rett syndrome conference will be on October 17-19th in Maastricht, The Netherlands.

ERSCM 2013 is an initiative of the GKC (Gouverneur Kremers Centrum), Stichting Terre (Dutch Rett Syndrome Foundation) and NRSV (Dutch Rett Syndrome Association) and is organized by the European Rett Syndrome Conference Maastricht Foundation.

ERSCM 2013 is actively supported by:

- **RSE (Rett Syndrome Europe)**
- **IRSF (International Rett Syndrome Foundation)**
- **ESRRA (European Scientific Rett Research Association)**
- **All European Rett Syndrome Parents Associations**

Early bird registration until July 1, 2013

Please visit their website for more details: http://www.europeanrettsyndromeconferencemaastricht.eu/

A record year in IRSF sponsored research: IRSF's cumulative research spend is $30M and two Phase 2 clinical trials are underway

In 2012, IRSF invested nearly $4M in our research program that funds peer-reviewed grants and contracts that give promise to advancing treatment strategies of Rett syndrome and other MECP2 disorders. IRSF is incredibly pleased with the strategy of the rigorous scientific review process mediated by members of the IRSF Scientific Review Board and followed by a programmatic review by the IRSF research team and IRSF's Research Advisory Committee. We commend all for their valiant efforts to identify the top-ranked applications that are deemed to have the highest scientific merit and meet IRSF's mission of accelerating the discovery and development of treatments for Rett syndrome.

Below we list all the projects funded in 2012. For more information on our funded projects, visit the following page: http://rettsyndrome.org/research-programs/grant-programs/funded-projects

2012 New Translational Research Awards

Angel Awards

- Daniel G. Glaze, MD, Baylor College of Medicine and Jeffrey Neul, MD PhD, Baylor College of Medicine and the Jan and Dan Duncan Neurological Research Institute
  A randomized, double-blind placebo controlled trial of NNZ-2566 (IGF-1{1-3}, glycyl-L-2-methylprolyl-L-glutamine acid) with open label extension in adults with Rett syndrome
- Walter Kaufmann, MD, Children's Hospital Boston
  A Phase 2b placebo-controlled cross-over study of rh-IGF1 (mecasermin [DNA] injection) for treatment of Rett syndrome and development of a novel biomarker of cortical function
- N. Carolyn Schanen, MD PhD, Nemours Biomedical Research and Jeffrey Neul, MD PhD, Baylor College of Medicine and the Jan and Dan Duncan Neurological Research Institute
  Nonsense suppression as a therapeutic approach to Rett syndrome

HeART Awards

- Daniela Brunner, PhD, PsychoGenics Inc.
  PPAR-sparing insulin sensitizers for Rett Syndrome
- Aleksandra Djukic, MD PhD, Tri-State Rett Syndrome Center, Montefiore Medical Center, Albert Einstein College of Medicine
  Language comprehension and processing in Rett syndrome: A pilot study of eye tracking
- 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
- Lee-Way Jin, MD PhD, Regents Of The University Of California - Davis
  Preclinical studies of allopregnanolone, a positive GABAA receptor modulator
- 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. Teriparitide on Osteoblast Function, Gene Expression and Bone Mass
- Dag Yasui, PhD, University Of California-Davis
  Investigation of CHRNA7 Ligands as Potential Rett Therapies

2012 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 IKβ 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+ Ion and Glutamate Homeostasis in Rett Syndrome
- Beth Stevens, PhD, Children's Hospital Boston
  Role of Microglia at Synapses in Rett Syndrome

2012 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 Analysis 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

2012 New Contracts

- Jeffrey Neul, MD PhD, The Jan and Dan Duncan Neurological Institute at Texas Children's Hospital and Baylor College of Medicine
  Creation of a DNA repository for Rett syndrome
- Alan Percy, MD, The University of Alabama at Birmingham
  Placebo-controlled trial of Lexapro (escitalopram) for anxiety in Rett Syndrome
- N. Carolyn Schanen, MD PhD, Nemours Biomedical Research
  Development of a Humanized Mouse Line by Knock-in

2012 Renewed Contracts

- Walter Kaufmann, MD, Children's Hospital Boston
  RettSearch Consortium's Clinical Coordinating Center
- Helen Leonard, MD, Telethon Institute for Child Health Research
  InterRett - Phenotype Database
- John Christodoulou, AM, Children's Hospital at Westmead
  RettBASE

HEALTHMATTERS

In response to questions into IRSF asking for information about hip issues, we invited orthopedic surgeon and IRSF orthopedic consultant Dr. John Killian to write for us on the subject, and we thank him for his time and experience. Dr. Killian's full article, which includes information on surgical interventions, can be accessed at http://www.rettsyndrome.org/understanding-rett-syndrome/living-with-rett-syndrome/symptom management/health-matters

The top-level message we want all parents and caregivers to hear is that not all children will have issues with their hips. These are a possible secondary issue that can develop over time as a result of apraxia, and imperfect motor and muscle control and coordination. Be aware, and involve your child's care team early on to make a plan for prevention and treatment.

Paige Nues
IRSF Director Family Support

This journey of a thousand miles begins with a single step -Lao Tzu

Rett Syndrome and Hip Problems

Growth and development of the hip is frequently abnormal in girls with Rett Syndrome. The hip joint is made up of a round femoral head, a spherical acetabulum, and "O-ring" or labrum. (Figure # 1). Four major muscle groups contribute to hip stability. The hip flexors raise the leg to walk, run and climb. The hip extensors stabilize the joint providing the ability to stand independently and run. The hip adductors and abductors assist with standing on just one leg at a time while walking. The full spectrum of abnormal muscle tone; hypotonia, spasticity, rigidity, and dystonia, exists in girls with Rett syndrome and can affect hip development.

The failure of the hip to develop normally is called dysplasia. Even without an x-ray, certain physical findings can indicate dysplasia: new leg length inequality, limitation of full abduction of the hips, or clunking sensation with hip abduction/adduction. An examination to determine whether any of these findings are present should be performed with the patient on a flat, firm surface. Leg length is measured from the front of the pelvis to the inside ankle bone. The position of the legs with respect to the torso is important because if the legs are swept to one side or the other, the measurement will be inaccurate. A difference of more than ¾ inch is considered abnormal or "positive". The next screening examination is performed with both hips flexed to 90 degrees. The hips are then allowed to "flop" outward or abduct to the maximal degree. If there is less than 45 degrees of hip abduction on either side, then the test is considered "positive". For the third screening examination, one hip is flexed to 90 degrees, and as the hip is abducted and adducted, the presence of a clunking sensation which may indicate the hip going in and out of the socket. If any of these tests are positive then an x-ray is indicated.

Of these screening examinations, limitation of full hip abduction is more likely to be associated with an abnormal secondary screening examination or x-ray. X-rays will be taken every 6-12 months to document development.

Parents, physical therapists, and orthopedic surgeons are aware that the growth and development of a normal hip is a prerequisite to regaining and maintaining ambulation in Rett Syndrome. Screening examinations should be performed yearly in the non ambulatory patient. X-rays should be performed when the physical examination is abnormal, tone in the lower extremities has dramatically changed, or a noticeable deterioration in ambulation has occurred.

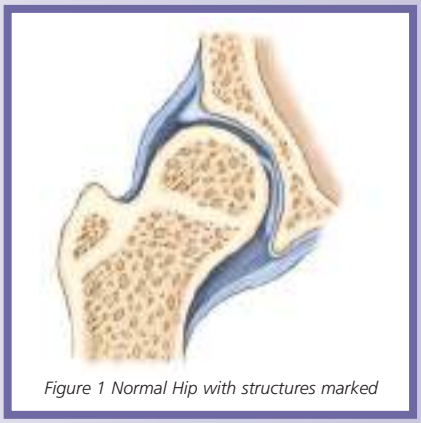


Figure 1 Normal Hip with structures marked

Message from the IRSF Family Support Team Members

At IRSF, we live for the moment when a child with Rett syndrome becomes healthier, happier, better educated or more openly embraced by her community. When parents feel secure in their parenting of our special children, feel hopeful and supported; when educators recognize their students learning potential; when physicians and therapists deliver better care plans; when a clinical trial has a long waitlist of eager trusting families; and when friends and families become bold fundraisers and advocates, then we know we are accomplishing our mission. These are the priceless gifts that only IRSF can give to the thousands of affected families who would otherwise be left adrift with nowhere to turn, without research hope on the horizon, raising a child misunderstood by many. Our every effort is to bring awareness and education about Rett syndrome to the community, to ask our support systems to do better by our kids every day, and impact the feelings of hope and inspiration across the world. It is our honor to wake up every day and deliver this support to the Rett syndrome community.

Sincerely,

Paige Nues and Jennifer Endres
IRSF Family Support
Team Members

POSSIBILITIES

Caitlin wins Jr. Miss Wheelchair of Massachusetts

Samantha participates in the Special Olympics

Emma bungee jumping

Sara meets Nemo the dolphin

Lillie hanging out with Elmo and Cookie Monster

Haley doing ballet

Eden holding drink box

Gia doing water therapy

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 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!

BELLA WITH GRANDMA GINGIE
I love Bella more than words can say. I love her precious little squeal; I love the sparkle in her gorgeous eyes but most of all I love to hear that special "giggle" that takes my breath away. She is my world!

GRACE WITH GRANDMOMMY
She is a precious girl and I love singing "You Are My Sunshine" to her.

CHELSIE WITH GRANDMA AND GRANDPA
Chelsie is the light of our lives! We call her "Chelsie Bug." She has been an inspiration to all around her but especially her grandparents!

EMMY WITH GRANDMA AND GRANDPA
Emmy loves it when Grandma sings, "I love you a bushel and a peck, a bushel and a peck and a hug around the neck." She smiles and leans in to give a "lovee."

ABBY WITH NANA AND GRANDPI
We love our Abby for her beautiful disposition and her great smile. She has added so much joy to our family.

ISABELLE WITH GRANDPA AND NANNA
Isabelle is our first grandchild and it was an instant bond of love with us from Day One. Although devasted by the recent news of Isabelle's diagnosis, she always will be our most precious and loving angel. Isabelle always has the most beautiful smile and we are ready to face RTT as a family team head-on.

LAUREN WITH OMA
What a blessing it is to be Oma to Lauren. Lauren fills my days with love and joy. Lauren is a sweet girl with a kind heart. Lauren is very sensitive about others and wants everyone to be happy. Everyone who knows Lauren loves her and is blessed by her wonderful outgoing personality.

REGIONAL REPRESENTATIVE PROGRAM

Here are messages from just a few of our dedicated Regional Reps! Contact yours today! Please visit http://www.rettsyndrome.org/family-support/support/regional-representatives

ANN-MARIE KINZLER • VA (eastern)
amkinzler@hotmail.com

In January this year I was asked by the nurse at my daughter Audrey's school if I would make a presentation to some nursing students and school staff on Rett syndrome. Of course I agreed! I want to raise awareness any chance I get. The thought that I would be able to give this information to future nurses excited me since they are really "first responders" in our health care. After the presentation, they asked great questions and were glad to have received the information. I have been asked to present again for another group of nursing students, as well as to speak at the nursing program at George Mason University. I really enjoy being a regional rep for IRSF and feel very privileged to be able to raise awareness for my daughter and all families living with Rett syndrome.

ARIANE MCMAHAN • CT
arianem@optonline.net

Hello! I would love a partner in crime! Is anyone interested in joining me as Co-Rep for the state of CT? Please contact me if you are—I'd love to hear from you!

CT Family Gathering

ASHLEY ALEXANDER • GA
Amfp786@yahoo.com

Hi Georgia families! We had so much fun at our family picnic, GA Strollathon and Blue Sky Girls events in 2012 and are looking forward to a very special 2013! It will be tough to top last years but, we will do our best. Please join us June 1st as we kick off the 3rd Annual Georgia Strollathon at our "Luau" family picnic! Be on the lookout for save-the-dates. You won't regret the lasting friendships you will gain by connecting with our fabulous families!

KANDY HALLSTROM • ID
kandylh@msn.com

Hello! We have a few people interested in helping to organize an ID Strollathon. Is anyone interested in taking the lead and being the Chair of the event? You would have a lot of assistance! The month of October would be a great time to hold it! Please contact me if you are interested.

LESLIE GREENFIELD • NJ
leslieg@optonline.net

Please consider joining the NJ and surrounding area families for a casual Rett Family Weekend April 19-20. There will be a short program to update everyone on the research then we will spend the day hanging out together at the indoor pool... having dinner as a group...staying overnight and having breakfast together. It is a great time to share the good and the bad...trade ideas...items... names of our favorite doctors, therapists etc. It is also wonderful for all the SIBLINGS to get together and see that they are not alone! Please contact me for more details! And don't forget about the NJ Natural History Study being held August 10-11.

PATTY MEVIS • WI
mmevis@sbcglobal.net

With spring upon us, many of us are in the planning stages of a variety of wonderful events happening across the great state of Wisconsin. I hope you'll mark your calendars to attend at least one event. They are great opportunities to connect with other families.

The 3rd Annual Cure for Keira, Drive for Hope golf outing is going to be held June 23rd, the annual Spike Rett Volleyball, Texas Hold-em and Beanbag Tournament is going to be held August 2nd & 3rd , and the 2nd Annual Green Bay Strollathon is going to be held September 28th! Please contact me for more information!

I also look forward to seeing some familiar and hopefully some new faces at the annual conference in June. I'd love to hear back from any families with ideas about where we could plan our next Wisconsin gathering. I invite you to contact me anytime!

MAUREEN CHARAMELLA • PA (EASTERN)
maureen.charamella@yahoo.com

The 2nd annual golf outing for Rett syndrome in memory of Rett dad, Mark Charamella, will be held in the fall of 2013. The date has not yet been set, but please contact me if you are interested in participating. Also, family picnic will be held in August—more details to come!

VANESSA PEACE • TX (NORTHEAST)
vanessa.ella.peace@gmail.com

We had a great 2012 with a family picnic in the spring and our first IRSF Strollathon on the last weekend of October. I am so thankful for all of you who participated. We had a good turnout at each event and all families benefitted from getting together and sharing our lives and stories. Not to mention how good it felt to have great support from Rett family and friends! We'd love to have even more families join us this year when we meet up again. Please contact me anytime for more information!

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Northeast TX Family Gathering

CHARLEY AND CRYSTAL WEAVER • IA
wwgang@iowatelecom.net

Hi Iowa families! It was so nice of all of you to join us for our 1st annual family gathering at the Blank Park Zoo. We are busy planning the 2013 gathering! Hope to see you there. Please contact us for more information!

IA Family Gathering

JOYCE OPINSKY • MO
joyce@gatewayshoes.com

Hey Missouri families! It was great to see those of you that attended our "Mom's Night Dinner" this winter. It was a really fun evening and hopefully the first of many! Be sure to mark your calendar for May 4th for the 6th Annual St. Louis Strollathon. It's a heart-warming, fun-filled morning to celebrate our girls as we stroll around beautiful Tilles Park. Hope to see you there!

SHERRI BRADY • CA (SOUTHERN)
Sbrady0457@charter.net

Hello SoCal Families! It was great seeing so many of you at our 10th Rett Family Picnic & Strollathon. We set new records with over 400 people in attendance and over $40k raised. This was in addition to many other great fundraising and awareness activities you all planned and carried out throughout the year...you guys are amazing. I hope 2013 is off to a good start for all of you and I look forward to seeing you at one of our upcoming events. Please don't hesitate to contact me if you have questions about local services, programs, the CHLA Rett Clinic, etc.!

SHELBY SWANN • IL
shelby.swann@yahoo.com

The Chicagoland Strollathon is quickly approaching! Planning is underway for our 2nd annual event to be held on May 19th. We know it is going to be a great event again this year. Princess Merida will be making an appearance for our Brave girls! I hope you will join us! Please contact me if you'd like to participate!

PETRINA PATE • VA (WESTERN)
petrina@roanokestrollathon.com

Hello VA families! I hope you have all had a great start to 2013! There are lots of exciting things going on in the "Rett World" with clinical trials and fundraisers galore! 2013 is going to be a great year! As far as VA events, be watching for more details, but for now, you can put 2 Roanoke events on your calendars: 1) June 1st-The 4th Annual Mix & Mingle for a CURE! 2) Sept. 14--The 9th Annual Roanoke Strollathon! Last year was our best year to date! We'd LOVE to have you make plans to get involved this year! Wherever you are located in the state, I hope you can find a way to participate in an event near you! I hope you all have a wonderfully blessed year and know that I am always here if there is anything you need even if it's just to talk!

NIKKI JOHNSON • FL
nikkij02@att.net

Hello to the local families in the NWFL area. Wanted to make sure you knew about the Strollathon we have planned coming up in April. Please email me for more information! Also, I would love to hear from you, and make sure I have your correct contact information. Please don't hesitate to contact me anytime! Look me up on Facebook too!

NC Family Gathering

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!

Keilly with her brother and sister

Tamara Lee with her sisters

Sorel with her brother

Jillian with her brothers

Lexie's sister

A Simile Poem by Bailey Larson

Emma

You are as sweet as an apple; a beautiful sister
You are as orange as autum; leaves in your hair
You are as strong as a fern; dew drops slipping off your leaves
You are as icy as water; your drips sliding down
You are as blue as night; comets shooting across your starry face
You are as quiet as a mouse; whiskers twitching
You are as exciting as a good book; I turn your pages like this poem

STROLLING INTO 2013

As the weather warms up, so do our volunteer efforts in planning local Strollathon events. Strollathons will be coming to at least 30 locations in 2013 and this is a great time to get involved. Taking the first step, whether registering a team for the first time or reaching out with interest in joining a committee or hosting an event in your community, can be nerve-wracking. It is **THAT** first step that opens you to great support, either from the local Strollathon chair and committee or from IRSF staff – and in many cases, both – and the opportunity to meet new families living with Rett syndrome in your local community!

Strollathons are fundraisers, friend-raisers, and family support rolled into one. Not only did our 2012 events set local records for dollars raised, but they also set records for children with Rett syndrome in attendance AND new family identification. Imagine the feeling of some of our local chairs in meeting families with daughters in their 20s who had previously never met another Rett syndrome family. This happened at a number of events!

2012 Georgia Strollers and Rollers

While Strollathons are community based, you don't have to be present on Stroll day to be part of the success. With online fundraising, you can raise money in honor of your loved one with Rett syndrome from anywhere in the country. Visit www.strollathon.org to view the current list of events and dates, and register for the event closest to you, the location where your loved one lives, or where you have family or friends who might also want to get involved to support your efforts. We look forward to seeing you in 2013!

Above (L to R) Kevin Black, Mark and Rachel Rohr, Kathryn and Luke Kissam

Clint Black onstage

Black Out Rett Syndrome

Each year, our board members take the lead in raising money to fund Rett syndrome research. In October 2012, Kathryn Kissam, IRSF board president hosted Black Out Rett Syndrome with a fantastic team of committee members and volunteers. With a live and silent auction, and appearances by Clint Black and Kevin Black, the event raised over $430,000 for Rett syndrome research. Families and friends in the Rett syndrome community from as far away as Texas and great local partners joined Kathryn for this festive evening.

www.rettsyndrome.org

Please visit the IRSF calendar to find events, large and small, happening in your area. If you don't see anything, consider hosting something yourself. IRSF staff can provide guidance and support in choosing your event and how to get started.

Contact Marcy Fritter at mfritter@rettsyndrome.org for more information.

Rett Racers with their inspirations after the Santa Barbara Marathon

Going the Distance

The Rett Racer program continues to grow. Racers have already signed up for more than 5 events in 2013 including marathons, half marathons, triathlons and more. In addition, Rett syndrome awareness will be part of the Boston marathon through the Rett Syndrome Association of Massachusetts.

If you are signed up for a race or plan to sign up, we can help you fundraise as you train. Online fundraising pages and resources will help you achieve success, and special incentive recognition is available beginning at $500 raised. Visit www.rettracers.org to sign up or for more information.

BEYOND THE RETT COMMUNITY

IRSF and Randstad USA are excited to announce our first formal organization-wide partnership in 2013. Randstad has long supported IRSF events in the Boston area, with involvement in other key events and markets. This new formal partnership will create opportunities to introduce Randstad staff in branches around the country to our families and volunteers in those communities. As the second largest staffing company in the nation and the world, Randstad is helping bring Rett syndrome awareness to hundreds of branches, over 2000 employees, and their local business partners.

Chicago area Randstad employees meet Kayla and her mom Kristin

Randstad has always had a philanthropic focus and, to reiterate its importance, rebranded their program as the Corporate Social Responsibility Outreach Initiative (CSR Initiative) three years ago. With this rebranding, the program has become more visible and robust. Randstad has identified four partners with a focus on children and health issues, including IRSF. Staff members are encouraged to participate in the CSR Initiative, from the time they complete their orientation, through days off to volunteer, payroll deductions, and contests to encourage engagement.

Randstad is helping bring Rett syndrome awareness to hundreds of branches, over 2000 employees, and their local business partners

Randstad's initial focus through this partnership is to engage local branches and staff members to participate in local Strollathon events. Randstad has set up an internal website to communicate information on upcoming local IRSF events and activities around the country and has a dedicated staff member facilitating the introductions and coordinating with IRSF for opportunities for branch staff to get involved.

Rett syndrome and IRSF have always been part of the Randstad family. Company president, Dan Foley, is close friends with a family whose daughter has Rett syndrome. Kristin Kelley, Randstad's Executive Vice President of Marketing, shared that the partnership with IRSF and the CSR Initiative as a whole will allow employees to feel like they've made a difference and to feel the sense of community that comes from families and friends rallying to support the individuals living with Rett syndrome and raising money to fund research to find a cure. Kristen left us with this message: "Thank you for allowing Randstad to get close and figure out how we can help make a difference."

Stay tuned for more information on Randstad partnership opportunities in your community and contact Marcy Fritter at mfritter@rettsyndrome.org or Jennifer Endres at jendres@rettsyndrome.org with questions.

memorials

The loss of every child is a loss for our entire community. Please join us in a moment of reflection for all of our beautiful children, and especially for these families who need our support during their difficult time. We promise to learn from each and every one them, remember them always, and allow their lessons to guide our efforts forward.

Kiera Dawn Delridge, 11/27/04-01/25/13. Kiera touched so many lives in the 8 short years we had the honor having her in our lives. She taught us the meaning of unconditional love. She taught her siblings about children with disabilities. In turn they all knew how to take care of most of Kiera's needs, and knew how to read her when she needed something. Always rewarded with bright shiny eyes, kisses, and a smile that would light up a room. Kiera is, was, and always will be our guiding light. Always live on in our hearts, taking all her smiles and love with us wherever we may go.

Kiera had a lot of pulmonary problems that led to pneumonia time and again. She fought long and hard. Collapsed lungs, extended hospital stays. She was tired. She is now in a much better place. Able to walk. Dance. Sing. And we know that one day we will all meet again. Until then, she will live forever in our hearts, and memories. We all love you and miss you greatly.

Danielle "Dani" Melissa Dolechek, lost her courageous and fiercely fought battle with Rett Syndrome on November 8, 2012 at the age of 18. She went straight to Heaven from home surrounded with love from her dad Bob, brother Derek and mom Elaine. She died from aspiration pneumonia, which had plagued her, non-stop, during the last 3 years of her life. She was the bravest little soul that we have ever known and our lives will never be the same without her. Although we are absolutely heartbroken, we know that she is finally out of pain and finally at peace, which is all we ever really wanted for her.

Dani was born on July 4, 1994 and we called her our "little firecracker". When she was little she walked independently, swam, rode horses, attended school, had play-dates and had a huge vocabulary of up to 300 words. Eating was by far her favorite activity, especially pizza, pasta, and "boogers" (burgers). She loved birthday parties and saying "birthday". She also had a bear that she named "Boe". He could say "I love you". So Dani would say "Boe" to everyone she cared about, every single day, because it was her way of saying "I love you". She would also ask us to always read her a "Book!" and her stunning green eyes always spoke where her words left off. It was heartbreaking when she stopped being able to eat, but she kept very busy with visiting with the use of her walker to her favorite places: Build-A-Bear, the Hallmark store and around the neighborhood.

The lives she touched were too many to mention. It showed at her service at St Paul's Catholic Church. Standing room only! She will be with us forever.

Natalie Ann Frohning, age 24, passed away at St. Luke's hospital in St. Louis, Mo., on June 20, from complications of pneumonia and hypo gamma globulin anemia. Natalie had an extraordinary determination to live and courageously fought in surviving many hospitalizations the last 8 years of her life, one being a 5-month hospitalization following spinal fusion surgery. The last 3 weeks of her life, she was surrounded by many friends, relatives, and medical professionals, who had grown to love her dearly for her what she had taught them about the preciousness of life.

This is how they said they remember Natalie. " She was a beautiful girl with a sunny personality, expressive eyes, and a glorious mop of dark brown hair, who brought sunshine wherever she went. "Her eyes were like the eyes of a fawn, God's precious little creation. Her eyes were so huge, full, bright, and her long eyelashes accentuated her eyes in a way that drew you into her world." "Her giggle was impossible to ignore, and her eyes would say, I think you are the most important person in the world, let's be friends." "She was such a fighter and taught me to work harder to push through obstacles and made us all more compassionate people."

Natalie would get extremely excited, when she recognized people that she knew. Her father, Fred, who passed away in 2004, said that Natalie was the happiest person he ever met. When he entered the room, she would greet him with the words, "Dad, Dad, my dad" and her mom, Linda, with "Mom, Mom, my mom" and others with excited words of "I see you, I see you."

We are all heartbroken that she is no longer with us, but are so grateful for the precious years that we all got to share with her. We feel that our precious angel, Natalie is sitting on a cloud watching over us. She is swinging her feet in the wind while listening to her favorite Elvis song, "the wonder of you" and telling us that she will send us sunny days.

Breanna Michelle Leatherwood, age 16, was a silent angel with a magnetic smile that immediately attracted your attention. Her quiet watchful eyes always shared her inner feeling. When her bright dazzling eyes and her smile erupted into laughter it coerced you to join in her chuckle. She truly enjoyed people and being the center of attention. She had a special way of letting you know she was always in command of her decisions whether happy, sad, content or discontent. Her lovely smile won the hearts of those she met in every aspect of her life.

Breanna endured many hospitalizations and surgeries and battled numerous gastrointestinal problems during her short life. Yet she championed each illness victoriously. The past eighteen months she endured nine months of hospitalizations. However she was able to enjoy her last nine months of life at home. Her health improved and enabled her to return to school as well as expand her use of the Dynavox communication device. Utilizing her eye gaze she was quite skilled at turning the pages to say what she needed to say. She rapidly acquired new skills and repeated return demonstration.

Breanna is extra special in our life for many reasons. Her existence as a silent angel truly touched and transformed our lives into the world of Rett syndrome. She was an instrumental Rett angel that initiated the development of the Southeastern Rett Syndrome Alliance. Through her life, she allowed many hearts to bond and connect each year during our annual conference and Alabama Rett Connect activities. Breanna was such a joy to have in our life. She has taught me so much in the short eight years I was privileged to share in her life. It is her strong spirit and memories of her laughter and pain that will enable us to continue her legacy within the southeastern region. Ironically it is her hardship, pain and laughter that enables us to become stronger advocates for improvements in several aspects of care, empowerment and awareness as we emerge from the heartfelt pain of moving forward without her visual presence. The spirit of Breanna will continue to build her legacy in the Rettworld.

Marilyn Archibald, Godmother to Heavenly Angel Breanna
President, Southeastern Rett Syndrome Alliance
Alabama Co- representative, IRSF

Emma Claire Reilly, 5 years old, was born September 15, 2007 in Maryland. She lived with her loving sister, Molly (12yrs); energetic brother, Seanie (9yrs); and parents, Beth & Sean Reilly. Emma loved watching "Yo Gabba Gabba", watching "The Fresh Beat Band", playing with her siblings and going to school. Her favorite was "talking" with the PODD communication book.

What a blessing Emma Claire was to all that knew her. How lucky she was to have a wonderful team of teachers and therapists that believed in her intelligence! Emma Claire was teaching people along the way.

Emma's journey with Rett Syndrome ended on November 11, 2012 due to aspiration from post-surgical complications. She is now able to sing and dance in Heaven, free of her earthly challenges (probably to the song, "I'm sexy and you know it"). We will miss Emma's beautiful eyes, her sweet giggles, silly raspberries, soft taps/hugs and her cuddles.

IRSF
INTERNATIONAL
RETT SYNDROME
FOUNDATION
4600 Devitt Drive
Cincinnati, OH 45246
www.rettsyndrome.org

this is the place for hope

IRSF 2013 Education & Awareness Conference

June 27-29, 2013
Midway, Utah

THE 29TH ANNUAL FAMILY CONFERENCE *The largest gathering and only one of its kind for parents, caregivers, extended family and professionals to come together to learn current care and advocacy strategies and the latest in research and clinical trials.*

In addition to world-class live presentations, lectures and workshops, attendees will also: interact with exhibitors; connect directly with the faces of IRSF; and attend the cherished Tribute Reception. Come to beautiful Utah, the place for education, awareness, identifying your strengths and those of your child, making new friends, and most of all, discovering through IRSF the place for hope!

Email Paige Nues, IRSF Conference Director, with any questions or ideas for sessions, Exhibitors, Sponsors, or Volunteer offers pnues@rettsyndrome.org

FEATURED THIS YEAR

- *Motivational keynotes*
- *Cutting edge information about the state of research and the currently funded Rett Syndrome Clinical Trials*
- *Communication experts and hands-on communication device workshops*
- *Expert sessions on medical issues, therapies, education, family foundations, financial and legal life planning*
- *Incredible information and support geared especially for all new parents of children with Rett syndrome and Related Disorders*
- *Largest targeted, tailored hands-on Exhibitor arena just for products and services that really matter to families, and make a difference for our children*

PRE- AND POST-CONFERENCE WORKSHOPS *For attendees interested in hands-on small group technology workshops, IRSF will partner with AAC experts* ***Judy Lariviere*** *and* ***Pati King deBaun*** *to host, for an additional fee, an iPad workshop on Thursday afternoon before conference officially begins, and a "Make and Take" page set workshop for high tech eye-gaze devices on Sunday morning. Please see website for additional information, bring your child's devices, and build this into your travel plans. These technologies will be examined throughout the conference sessions on Communication, and IRSF is pleased to offer these additional hands-on workshops with our AAC specialists and technology partners to maximize your time in Utah.*

NATIONAL ABILITY CENTER DAY PROGRAM FOR THOSE WITH RETT SYNDROME AND RELATED DISORDERS *IRSF is pleased to partner with the National Ability Center in Park City, Utah to offer a fun-filled day program for our children with Rett syndrome on Friday and Saturday for a nominal fee. See website soon for more details. IRSF will not be offering onsite Respite. We encourage you to register for this program if your child is traveling with you to conference and you have a caregiver, spouse, grandparent or mature sibling who can accompany her and attend to her personal care needs throughout the day. This will be a wonderful outing for her!*

THE IRSF 29TH ANNUAL FAMILY CONFERENCE JUNE 27 – 29, 2013 IN MIDWAY, UTAH

****Early Bird Rate: $285 per person for Full Conference Registration and $180 for Single Day Registration Before May 27th.***

****Note: Contact The Zermatt Resort or their sister property The Homestead (right across the street!) for hotel rooms. Visit www.rettsyndrome.org or call The Zermatt Resort at 1-866-937-6288 and reference the IRSF Annual Family Conference for negotiated rates starting at $149/night (Zermatt) and $129/night (The Homestead). Salt Lake City International Airport is recommended for air travel. Refer to the website for more detailed conference information.***