Dear Rett syndrome community,

As we prepare for the 2012 holiday season, the IRSF board & staff would like to wish each and every one of you, a very happy, healthy and joyous Thanksgiving. As we look back on this year there is much to be grateful for.

We are thankful to the generosity of Ms. Judi Rees and the Rachel Ames Rohr Foundation as they have given IRSF the opportunity to double the impact of your year-end contribution. All first-time and increased gifts, totaling up to $100,000, will be matched dollar per dollar. Please click here to contribute today to make an even greater difference. Thank you.

IRSF is also grateful for all of the amazing individuals who did so much to make our month special. Over 25 incredible events hosted by families and supporters of IRSF raised spirits and critical funds in October, including the signature IRSF Strollathons held in Southern Florida, Grand Rapids, and Fairfax among others. The PSA, Governor Proclamations, Blue Sky Girls events, the dedicated Rett Racers as well as events like the Reeling it in for Rett Fishing Tournament, Spending Time Ending Rett Concert, The Black & White Affair and the Black Out Rett Syndrome Concert are just a few examples of the many ways that you helped spread awareness during our month.

We are thankful for the hard work and dedication of our funded researchers who are bringing forth two clinical trials that were funded by IRSF this year. While IRSF originally announced that the trials would have begun in the Fall of 2012, there have been delays due to University Regulatory oversight. Therefore, we give you a quick update on the status of these trials.

Baylor College of Medicine and Neuren Pharmaceuticals will bring the first clinical trial using a new drug designed by Neuren for adults diagnosed with Rett syndrome. With IRSF funding, they are ready to move forward with a Phase I and Phase II clinical study of an IGF-1 derived tripeptide called NNZ-2566. Upon FDA approval of the IND application, the trial is slated to begin in the early part of 2013.

Boston Children’s Hospital will conduct Phase 2 of the IRSF funded IGF-1 clinical trial with 30 girls by the end of 2012. The support of the Rett syndrome community helped put IRSF in the position to provide an ANGEL grant of $600,000 needed for this trial.

Lastly, IRSF is grateful for each of you. Because of you, our Board members, volunteers, representatives, our community, IRSF has made strides in our mission to accelerate research, raise awareness and funds all while supporting families in need. Your continued volunteerism and commitment extends our reach, strengthens our support, and creates a bonded, unified community. We have much to look forward to, our belief is strengthened every day by the commitment of families and the great work of our scientists that real change for families affected by Rett syndrome is in our future.

Happy Thanksgiving!
Events

Local Houston-area Roller Derby Team, aka Breanna’s Brigade, led the way at the Texas Strollathon in The Woodlands.

Thank you for your support in building another successful Strollathon season! With 21 Strollathons in 24 locations, we have raised over $1 million dollars in 2012. This included one Strollathon celebrating its ninth year and six more celebrating their first. These events are only successful because of you, our amazing volunteers who lead and participate in them, recruit family and friends to fundraise with you, and who share your personal stories of Rett syndrome with your communities. It is because of your efforts that the Foundation continues to be able to support important Rett syndrome research and clinical trials! Online fundraising pages for Strollathons in September and October will remain open through the end of the year and mailed donations will continue to be accepted for all 2012 events.

Conversations have begun with new and returning Strollathon Chairs for the 2013 season. If you are interested in learning more about bringing a Strollathon to your community, please contact Marcy.

Rett Racers

Congratulations to Nicky Meehan, Amy Drysdale, Lissy Binder, Meir Toshav and Jason Weissman for raising over $15,000 as they trained to complete the NYC Marathon. We share in their disappointment that Superstorm Sandy forced the cancellation of the event, but we know that their desire to cross the finish line for Rett syndrome remains strong! Nicky and Amy still ran the 26.2 miles on Sunday morning – that is dedication! All of their online fundraising pages are still accepting donations through the end of the year. Click on an individual name to support that Rett Racer.

The final Rett Racer weekend of 2012 saw Chris Kolendrianos complete the Santa Barbara International Marathon and a team of Racers
Superstorm Sandy and a canceled marathon didn’t stop Nicky Meehan from completing her 30 miles!

Featured Family Fundraiser

October was Rett Syndrome Awareness Month and we launched the Black Out Rett Syndrome campaign which culminated in the fantastic Clint Black concert event in Baton Rouge, LA hosted by IRSF Board Chair, Kathryni Kissam.

This was not the only Black family event during the month of October. For the last eight years, Kevin Black and his friend Dave Clements have hosted a variety of events in Texas in honor and memory of Kevin’s daughter Cortney. This year’s Spending Time Ending Rett events included a fishing tournament, a golf tournament, and a crowd-pleasing concert at the Dosey Doe “Big Barn” in The Woodlands. Kevin was joined on the stage by his son Coleton, the legendary Gary P. Nunn, and Jamie Richards. These events bridge the gap between celebrating our local Rett families and building awareness within the community through Kevin’s draw as a musician. In addition to sponsorships and ticket/participation purchases, each activity also included a live and silent auction, and raffles.

Congratulations on another successful year of events and thank you for your continued support, Kevin and Dave!

Research

Mutant Mice Reveals Functional Deficits in Forebrain Circuits, Including Key Nodes in the Default Mode Network, that are Reversed with Ketamine Treatment


Abstract

Article

Growth failure and outcome in Rett syndrome: Specific growth references


Abstract

Article

Activity-dependent BDNF release and TRPC signaling is impaired in hippocampal neurons of Mecp2 mutant mice

Li W, Calfa G, Larimore J, Pozzo-Miller L

Abstract

Article

Female Mecp2+-/- mice display robust behavioral deficits on two different genetic backgrounds providing a framework for pre-clinical studies


Abstract

Article

Regional MeCP2 expression levels in the female MeCP2-deficient mouse brain correlate with specific behavioral impairments

Wither RG, Lang M, Zhang L, Eubanks JH

Abstract

Article

A Critical and Cell-Autonomous Role for MeCP2 in Synaptic Scaling Up

Blackman MP, Djukic B, Nelson SB, Turrigiano GG

Abstract

Article

Improved Survival and Reduced Phenotypic Severity Following AAV9/MECP2 Gene Transfer to Neonatal and Juvenile Male Mecp2 Knockout Mice


Abstract

Article

Phosphorylation of MeCP2 at Ser421 Contributes to Chronic Antidepressant Action

Hutchinson AN, Deng JV, Cohen S, West AE

Abstract

Article

LINE-1 Retrotransposition in the Nervous System

Thomas CA, Paquola AC, Muotri AR

Abstract

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

Contributed by: Nurit Ballas, PhD

It is well established that mutations in the X-linked gene Methyl-CpG-binding protein 2 (Mecp2) cause Rett syndrome (RTT) at a specific postnatal stage. Although the mutation is present from birth, RTT syndrome in humans and mouse models is overtly manifest postnatally. The latency period to symptom initiation is about 12-18 months in human RTT girls, and 6-8 months in female afflicted mice, and approximately 5 weeks in the male mice. Recently, the Zoghbi, Bird and Ballas groups showed that loss of MeCP2 postnatally, even at adult stage, gave rise to the same spectrum of severe symptoms as when MeCP2 was lost at birth, suggesting 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.

Read more...

Abstract

Drug Reverses Abnormal Brain Function in Rett Syndrome Mice

A few weeks ago, a promising new study was published in the Journal of Neurosciences by Dr. David Katz and his colleagues at Case Western Reserve University that demonstrated that in a mouse model of Rett syndrome they were able to reverse abnormalities in brain activity and improve neurological function by treating animals with an FDA-approved anesthesia drug, ketamine.

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.

Read more...

Abstract

Natural History Study Pictures!

IRSF needs Natural History Study pictures and quotes about why the NHS is so important to you, the participating families. We especially need pictures of your child with our wonderful doctors, or tell us about any and all great “magical”...
moments with your Investigator, or maybe about the friendships and learnings you gain from being with other families. Group pictures are needed too! Why does it matter to you to give to research in this way? Please email directly to Paige Nues at pnues@rettsyndrome.org before the end of November, and we will get a photo release form out to you.

Featured App: NHGRI Talking Glossary of Genetic Terms
Talking Glossary of Genetic Terms features more than 250 common genetic terms pronounced and explained in an easy-to-understand way by leading scientists and professionals at the National Human Genome Research Institute (NHGRI).

State Facebook Pages!
Some of your Regional Representatives have created state Facebook pages! The mission of these pages is to connect the families of each state impacted by Rett syndrome in a place where many of you already are – FACEBOOK! Utilize your state’s page to chat about Rett syndrome issues, share in the excitement of IRSF events and connect with individuals near YOU.

Search for your state’s page today!

The Rett Gazette
Don’t receive the Rett Gazette? Check it out online! Sign up to receive the Rett Gazette

The Spirit of Giving!
Use our IRSF App, a safe, free and revolutionary approach to online giving. Every purchase and search you make online can generate money for IRSF. Please download The IRSF APP. We can now raise money from big online stores such as Amazon, Skype, eBay and thousands of others at no cost to you. Simply download the IRSF App using Firefox, Internet Explorer or Chrome and continue searching and shopping as usual. You can choose to install the app as a toolbar or as a small icon . It only takes a minute to download. If you do not like the app, it takes one click to uninstall.

Help us spread the word among your family and friends.

In September of 2011, a workshop focused on the state of the art in animal studies of Rett syndrome (RTT) was convened by the National Institute of Neurological Disorders and Stroke (NINDS), the Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD), the International Rett Syndrome Foundation (IRSF) and the Rett Syndrome Research Trust (RSRT). 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. On October 31, the outcomes of this workshop were published as an Open Access review article, authored by several workshop participants, in Disease Models & Mechanisms (DMM). As explained in this article, 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.

Read more...

Abstract
Article

Happenings

Research Webinar
Spare time over the holidays? Be sure to watch IRSF’s webinar recording from Oct. 23rd of Jennifer Larimore, PhD, Agnes Scott College and IRSF funded investigator, presenting “From Mind to Molecules: Neurobiology of RTT.” Fresh from the annual Society for Neurosciences meeting, we heard the latest news in easy to understand terms. Fact: did you know that in 2 minutes, your brain consumes more energy than your legs need to run a marathon? Nourish your brain with this webinar, and nourish those brains of our children with Rett syndrome by exposing them to an enriched and varied environment every day!

Read more...

IRSF FAMILY EDUCATION AND AWARENESS CONFERENCE – SAVE THE DATE!
The 29th annual conference will be in Midway, Utah, from June 27 – 29, 2013 (Thursday evening – Saturday evening). Start planning now. This conference is THE place to meet with the experts, make new friends and learn all things Rett-related: school and communication, therapies, medical issues, life planning, and everything current about research and clinical trials. Join us! Online registration will open end of January 2013.

IRSF Holiday Cards

Happy Thanksgiving
The Combined Federal Campaign
CFC Code Number: 11046
If you have a fair happening near you, contact the office at 1-800-818-7388 for materials.

Donations
Please send to P.O. Box 706143 Cincinnati, OH 45270-6143.

Matching Gift Program
Check if your company has a matching gift program. Send forms to lhayden@rettsyndrome.org or by fax at 513-874-2520.

Mail
Please send to 4600 Devitt Drive Cincinnati, OH 45246.

"Success is not final, failure is not fatal: it is the courage to continue that counts."
~ Winston Churchill

Since IRSF has received such an overwhelming response for this year’s Holiday Card design, we have decided to extend the deadline into early December! Get your Holiday Cards today & Keep Rett syndrome in mind while wishing your family and friends Happy Holidays!

$15 per pack
Each pack contains 12 cards and envelopes (Two pack order minimum)

Click here to order yours today!

Note: Due to fixed shipping and handling fees, we will be unable to accept international orders.

IRSF is the world’s leading private funder of basic, translational and clinical Rett syndrome research, funding over $28M in high-quality, peer-reviewed research grants and programs to date. Annually, IRSF hosts the world’s largest gathering of global Rett researchers and clinicians to establish research direction and priorities while exchanging ideas and the most recent information. IRSF is the most comprehensive non-profit organization dedicated to providing thorough and accurate information about Rett syndrome, offering informational and emotional family support and stimulating research aimed at accelerating treatments and a cure for Rett syndrome and related disorders. IRSF has earned Charity Navigator’s most prestigious 4-star rating. To learn more about IRSF and Rett syndrome, visit www.rettsyndrome.org or call IRSF at 1-800-818-7388 (RETT).

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