

IRSFflash:10.14.2011

October

14

[View Web Version](#)

[Share](#)

[Facebook](#)

[Twitter](#)

rettsyndrome.org

Right click and "Save As" to download image

This month is dedicated to raising awareness and it will only happen with ACTION!

Rett Syndrome Awareness Month is the perfect time to take action in the fight for a cure for Rett syndrome. Your action today will help to raise awareness, fund crucial research and eventually create a world without Rett syndrome.

Let us be even more ambitious this October Awareness Month than last. Off the heels of the launch of the NEW Research to Reality Clint Black PSA, now is our time to take action. You have so many different avenues readily available to assist you in the action of your choice. Give a gift, get involved with the PSA, wear purple, take part in an IRSF fundraiser and share our Facebook daily Rett Facts and October Awareness posts.

The IRSF leadership is supporting an ambitious three year Strategic Research Plan which will push the boundaries of basic research and successfully build our translational research program to bring two, possibly more, compounds to

eventual new clinical trials.

The IRSF board and staff is committed to raising funds and awareness for Rett syndrome this October and supporting you in your fundraising and awareness raising efforts. With your support, we will succeed; together we hope, discover and achieve.

Stephen Bajardi

Executive Director

IRSF

"Notable Honors Awarded to Rett Investigators"

Rudolf Jaenisch received the National Medal of Science

This past month President Barack Obama announced that Rudolf Jaenisch, MD from the Whitehead Institute with the nation's highest scientific honor- the National Medal of Science. This award recognizes his outstanding contributions to knowledge in the biological sciences that pertain to how genetic information is variably expressed through epigenetics.

[Read More...](#)

NIH Director's Early Independence Award given to Rodney Samaco

Rodney Samaco, PhD from the Baylor College of Medicine is an up-and-coming, talented, young Rett investigator, who IRSF would like to congratulate for receiving the NIH Director's Early Independence Award for his project entitled "The Genetic and Neuroanatomical Origin of Social Behavior".

[Read More...](#)

A Rett Workshop focused on "Setting Priorities for Therapy Development in Rett Syndrome"

The National Institute of Neurological Disease and Stroke (NINDS) of the National Institutes of Health (NIH) sponsored a workshop focused on "Setting Priorities for Therapy Development in Rett Syndrome" that occurred on September 25-27 in Bethesda, MD. IRSF and RSRT also co-sponsored this workshop that brought together basic research scientists, clinicians, representatives from the pharmaceutical and biotech industries, and officials from the FDA and the NIH.

[Read More...](#)

Join the Webinar: Inside the IGF-1 Clinical Trial for Rett Syndrome

The International Rett Syndrome Foundation, in conjunction with Boston Children's Hospital Rett Syndrome Program, invites you to an exclusive inside look at the IGF-1 Clinical Trial for Rett Syndrome with Principal Investigator Dr. Omar Khwaja and team. Please join us on October 18th to learn more about how this clinical trial came about and a current trial status update.

WEBINAR: How Do Clinical Trials Happen? An Inside Look at the IGF-1 Clinical Trial for Rett Syndrome with Principal Investigator Omar Khwaja, MD, PhD, Director Rett Syndrome Program, Children's Hospital Boston

Tuesday, 18th October, 2011

4pm eastern / 3pm central / 2pm mountain / 1pm pacific

** please access webinar or phone conference 5 minutes before the hour so we may begin on time **

[Read More...](#)

Investigator Spotlight: Liang Zhang, MD, PhD, University Health Network, Toronto Western Research Institute

With the many activities of this past summer, it is a pleasure to welcome the fall and continue with our Investigator Spotlight series. For this month's installment, it is a pleasure to focus on Dr. Liang Zhang from the University Health Network at the Toronto Western Research Institute. Dr. Zhang is an active participant in IRSF's research program, as he recently presented his work at the 12th Annual Rett syndrome Symposium and has been funded by IRSF's translational HeART grant mechanism since 2010. His project titled "Evaluating Carbonic Anhydrase Inhibitors as Potential Treatments for Rett Syndrome" aims to study an anti-convulsive drug (acetazolamide) in its ability to improve the neural and behavioral symptoms in MeCP2-deficient mice.

[Read More...](#)

NORD Sees Progress for Rare Disease Community in Newly-Released PDUFA Document

Peter L. Saltonstall, president and CEO of the National Organization for Rare Disorders (NORD) today issued the following statement of support for provisions related to rare diseases in the Food and Drug Administration's newly-released "PDUFA V" document:

"The FDA's newly-released PDUFA V document entitled 'Advancing Development of Drugs for Rare Diseases' contains elements for which NORD has been advocating for some time. We are especially delighted that this document calls for an additional five staff members in the Rare Disease Program in the FDA Center for Drug Evaluation and Research (CDER), and the creation of a liaison within the Biologics Center. The document reflects a clear recognition that drugs for rare diseases warrant special consideration and special staff training. On behalf of the patient community, I want to thank the FDA for working with us on these advances in the regulatory framework, and we look forward to continued collaboration."

.....

The draft document has been posted on the FDA website. It outlines the current status of the documentation for the reauthorization of the Prescription Drug User Fee Act.

Established in 1983, NORD is a nonprofit organization representing the nearly 30 million Americans who have rare diseases. It provides advocacy, education, research, and patient assistance programs on behalf of all Americans affected by rare diseases.

Parents Making a Difference

Popping into the grocery store for a gallon of milk or a loaf of bread isn't an option for Drew Ann Long. Instead, the Alabaster woman must carefully plan her shopping trips, waiting until her husband is home or arranging for a baby sitter to stay with her daughter. Ten-year-old Caroline has Rett syndrome, a neurodevelopmental disorder that causes multiple disabilities, and it's difficult for Long to navigate store aisles with her in tow.

That's a common experience among moms of special needs children, Long said, which is why she created a shopping cart to make the task easier.

[Read More...](#)

Blue Sky Girls!

Please learn more and become a Location Site Chair to join and support Dr. Sasha Djukic, Director, Tri-State Rett Center at Montefiore Rett Center in New York, in her efforts to create a ground-breaking, unified Rett syndrome awareness event around the world on October 15, 2011.

[Click here to download the flyer](#)

Featured Angel: Sejal Shah

Hi! My name is Sejal, and I am 5 years old. I was born in Williamsville, New York, and I was diagnosed with Rett syndrome when I was 2. I have been through some tough times, but now I'm doing great. I have learned to walk short distances, I am learning to communicate with my eyes, I am starting to learn my letters and numbers, and I am happy, giggly, and healthy almost all the time. And in the fall, I'm going to start Kindergarten -- I can't wait!

[Learn more about Sejal and her family...](#)

[How are you getting involved during October?](#)

October is Rett syndrome awareness month – YOUR HELP IS OUR HOPE! There are so many ways to get involved. Here are just a few to get your creative juices flowing!

[Participate in an existing event near you!](#)

October is the busiest month for volunteers and donors at IRSF. All over the country you can find unique, fun, and exciting fundraisers to take part in!

[To find an event near you click here to view our website calendar!](#)

[Create your own fundraiser!](#)

[Click here to download the A-Z list of fundraising ideas.](#) Turn your favorite activity into a fundraiser. IRSF has the tools to help make your event a success – INCLUDING an Event Starter Kit! Visit the Fundraising section or contact Jenni Grammer at jgrammer@rettsyndrome.org if you need assistance with planning your event or would like it listed on our website calendar.

[Each Month IRSF will Provide you with a Fundraising Idea!](#)

These ideas can be used throughout the year to keep the fundraising going and keep your community involved! Many of the Fundraisers we are going to share can be put together fairly quickly and are easy to do!

October is Rett syndrome awareness month so what better time to host a HER Night event! HER (Help End Rett) Night events are 100% customizable; you can host a dinner at your home, a back yard barbeque and 50/50 raffle, Sleepover party, the ideas are endless! Click [here](#) for a one page fact sheet with ideas on how to host your HER Night fundraiser. IRSF can provide you with the logo and sample invitations to invite your friends and family to your unique HER Night event.

For years IRSF supporters have enjoyed getting invitations to have a “cup of tea” in honor of girls and women all over the country and even the world! The Phantom Tea is for those that enjoy direct mail fundraisers. To download the order form please click [here](#) and fax or email to jgrammer@rettsyndrome.org.

For those of you who would rather do your fundraising online we have the IRSF Care and Cure Café via FirstGiving! Go to www.firstgiving.com/irsfcareandcure to create your very own fundraising page! It's completely customizable and easy! You can use this all year round too – in lieu of birthday presents, holiday gifts and other special occasions!

*If you have an event coming up and would like IRSF to promote via our website Calendar (link to attached calendar form [here](#)) please fill out this form and return email to jgrammer@rettsyndrome.org.

[Join a Support Network](#)

[Find your Regional Rep](#)

[Shop](#)

Past Issues

Zumbathon

Queen Creek Horse Expo - Awareness Event

Blue Sky Girls

6th Annual Fairfax Strollathon

An Inside Look At The IGF-1 Trial

IRSF 11th Annual Festival Of Food & Wine

1st Annual Reeling It In For Rett

Rendezvous For Rett

2nd Annual So Cal Strollathon And Picnic

2nd Annual South Florida Strollathon

Recent Publications

Synaptic microcircuit dysfunction in genetic models of neurodevelopmental disorders: focus on Mecp2 and Met.

Shepherd GM, Katz DM.

[Abstract](#)

[Article](#)

De novo Interstitial Triplication of MECP2 in a Girl with Neurodevelopmental Disorder and Random X Chromosome Inactivation.

Mayo S, Monfort S, Roselló M, Orellana C, Oltra S, Armstrong J, Català V, Martínez F.

[Abstract](#)

[Article](#)

Intrathecal 5-azacytidine inhibits global DNA methylation and methyl- CpG-binding protein 2 expression and alleviates neuropathic pain in rats following chronic constriction injury.

Wang Y, Liu C, Guo QL, Yan JQ, Zhu XY, Huang CS, Zou WY.

Abstract

Article

F2-Dihomo-isoprostanes as potential early biomarkers of lipid oxidative damage in Rett syndrome.

De Felice C, Signorini C, Durand T, Oger C, Guy A, Bultel-Ponce V, Galano JM, Ciccoli L, Leoncini S, D'Esposito M, Filosa S, Pecorelli A, Valacchi G, Hayek J.

Abstract

Article

MeCP2 and Rett syndrome: reversibility and potential avenues for therapy.

Gadalla KK, Bailey ME, Cobb SR.

Abstract

Article

Mecp2 Truncation in Male Mice Promotes Affiliative Social Behavior.

Pearson BL, Defensor EB, Pobbe RL, Yamamoto LH, Bolivar VJ, Blanchard DC, Blanchard RJ.

Abstract

Article

Temporal and Regional Alterations in NMDA Receptor Expression in Mecp2-Null Mice.

Blue ME, Kaufmann WE, Bressler J, Eyring C, O'driscoll C, Naidu S, Johnston MV.

[Abstract](#)

[Article](#)

Oxidative stress in Rett syndrome: Natural history, genotype, and variants.

Leoncini S, De Felice C, Signorini C, Pecorelli A, Durand T, Valacchi G, Ciccoli L, Hayek J.

[Abstract](#)

[Article](#)

Loss of mecp2 in substantia nigra dopamine neurons compromises the nigrostriatal pathway.

Gantz SC, Ford CP, Neve KA, Williams JT.

[Abstract](#)

[Article](#)

Two new Rett syndrome families and review of the literature: expanding the knowledge of MECP2 frameshift mutations.

Ravn K, Roende G, Duno M, Fuglsang K, Eiklid KL, Tümer Z, Nielsen JB, Skjeldal OH.

[Abstract](#)

[Article](#)

De novo deletion in MECP2 in a monozygotic twin pair: a case report.

Mittal K, Kabra M, Juyal R, Bk T.

[Abstract](#)

[Article](#)

Folinic Acid Supplementation in Rett Syndrome Patients Does Not Influence the Course of the Disease: A Randomized Study.

Hagebeuk EE, Duran M, Koelman JH, Abeling NG, Vyth A, Poll-The BT.

[Abstract](#)

[Article](#)

Affinity-based enrichment strategies to assay methyl-CpG binding activity and DNA methylation in early *Xenopus* embryos.

Bogdanovi O, Veenstra GJ.

[Abstract](#)

[Article](#)

A miRNA-Based System for Selecting and Maintaining the Pluripotent State in Human Induced Pluripotent Stem Cells.

Di Stefano B, Maffioletti SM, Gentner B, Ungaro F, Schira G, Naldini L, Broccoli V.

[Abstract](#)

Article

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.

Find out if your state has a page!

The Rett Gazette

Don't receive the Rett Gazette?

Check it out online!

Sign up to receive the Rett Gazette

Write a Rett Review

If you love helping us fight for a cure for Rett syndrome then tell the world! You have an exciting opportunity to help us make an even bigger impact in our community. Charity Navigator has partnered with GreatNonprofits to enable people to share their stories about nonprofits that have touched their lives!

Please help us raise visibility and support by posting a review. All reviews will be visible to potential donors and volunteers.

It only takes a few minutes! Go now!

Henhouse Coffee Partners with Rett Syndrome!

You can now buy your premium coffee AND contribute to finding a cure for Rett syndrome without ever leaving your home!

Visit www.Henhousecoffee.com, browse the website to learn more about Henhouse, the art of coffee roasting and discover which brew is right for you, choose your blend, click "Buy Online" and make your selection. BE SURE to select IRSF under "Support a Partner Organization". The coffee will be shipped directly to your door and IRSF will receive a donation of \$3.50 per 12 oz. bag.

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.

The Combined Federal Campaign

CFC Code Number: 11046

The 2011 CFC campaign continues through November. 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.

"When we change the way we look at things, things change the way they look."

~ Lee J. Colan

IRSF is the world's leading private funder of basic, translational and clinical Rett syndrome research, funding over \$24M 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).

Designed by Songswift: global village solutions

© 2011 All Rights Reserved