

## IRSFlash: Tell a Friend

Tell a Friend

{mosloadposition recommend}

Is this email not displaying correctly? Try the web version.

December

23

[View Web Version](#)

[Share](#)

[Facebook](#)

[Twitter](#)

[rettsyndrome.org](http://rettsyndrome.org)

Dear Rett Syndrome Community,

On behalf of the board & staff of The International Rett Syndrome Foundation I thank you for raising funds and donating your time, talent and expertise to make this year a successful one for IRSF and the Rett syndrome community. It is

because of all of you IRSF was able to fund 26 grants for nearly \$2.2 M for Rett syndrome research in 2011. These high quality grants were selected from a total of 81 applications with a cumulative request of \$8.4M for basic and translational research funding. Together, we are advancing science and making a difference in the lives of many families. As we pause during the holiday period we can look forward to next year with great optimism and hope. The light in front of us is getting brighter!

All of us extend our deepest wishes for a happy and healthy holiday season to you and your family, thank you for being a part of the family and thank you for supporting IRSF in the fight to develop treatments and find a cure for Rett syndrome.

Stephen Bajardi

Executive Director

IRSF

Save the Date: World Rett Syndrome Congress

The 7th World Rett Syndrome Congress, hosted by IRSF will be held in New Orleans from June 22 - 26, 2012. This unique event begins with IRSF's Family and Professional Education track June 22 - 24. The Scientific Symposium will be chaired by Huda Y. Zoghbi, M.D. (Duncan Neurological Research Institute, Baylor College of Medicine) and Gail Mandel, Ph.D. (Vollum Institute, Oregon Health & Science University) June 24 - 26, and the Pre-Clinical and Translational Research Symposium will be chaired by Jeffrey Neul, M.D., Ph.D. (Duncan Neurological Research Institute, Baylor College of Medicine) on June 24. The World Congress is hosted once every four years, and 2012 marks the first time it will be held in the United States.

Interested sponsors and exhibitors for this event should contact IRSF immediately at [admin@rettsyndrome.org](mailto:admin@rettsyndrome.org), with "World Congress" in the subject line.

Registration will open early 2012.

Location: Intercontinental Hotel, New Orleans, LA. A custom link for negotiated World Congress room rates will be shared soon

Sessions for families, caregivers and professionals: Friday through Sunday, June 22 – 24, 2012

Meetings for scientists: Sunday through Tuesday, June 24 – 26, 2012

IRSF awards over \$1.5 million for basic and translational research for Rett syndrome

New awards bring IRSF's cumulative research spend to over \$26 million

Cincinnati, (OH) - The International Rett Syndrome Foundation (IRSF) announced today that it is awarding over \$1.5M to support 18 new grants designed to study a variety of diverse topics from basic science and disease pathology to developing treatments and outcome measures for Rett syndrome. IRSF is the world's largest private source of funding for biomedical and clinical Rett syndrome research with the cumulative total of research spending of over \$26M since 1998. It is the mission of IRSF to invest in these high quality, peer-reviewed basic and translational research grants to advance research towards new treatments for Rett syndrome.

[Read More...](#)

Investigator Spotlight: Aleksandra Djukic, MD, PhD, Tri-State Rett Syndrome Center, Montefiore Medical Center, Albert Einstein College of Medicine

To end this year, it is with great pleasure to feature Dr. Aleksandra Djukic in this installment of the Investigator Spotlight series. Dr. Djukic is the Director of the Rett Syndrome Center at the Montefiore Medical Center and is also appointed as an Associate Professor of Neurology at Albert Einstein College of Medicine of Yeshiva University. She attended the University of Belgrade in Yugoslavia-Serbia for both medical and graduate school, received training as a Pediatric Resident and Neurology Fellow at the Albert Einstein College of Medicine in Bronx, NY, and has an extensive background as a neurologist and neuropsychologist.

[Read More...](#)

### Pathogenesis of Lethal Cardiac Arrhythmias in MeCP2 Mutant Mice: Implication for Therapy in Rett Syndrome

Although many people with Rett syndrome live long lives, up to a quarter of all deaths in Rett syndrome are sudden and unexpected. Dr. Neul and his colleagues explored the hypothesis that these deaths are due to cardiac dysfunction. Using data collected in the Rett Syndrome Natural History Study, they found that nearly 20% of those with Rett syndrome exhibit changes in the way their hearts conduct electricity. This type of problem can make the heart suddenly start beating very fast (tachycardia), which can lead to an inefficient distribution of blood and result in sudden death.

[Read More...](#)

[Abstract](#)

[Article](#)

### A new mouse model for the human Rett syndrome mutation MeCP2 T158A

The Methyl-CpG-Binding Protein 2 (MECP2) gene was identified by Dr. Adrian Bird and his colleagues as a gene that encodes for the multi-functional MecP2 protein that regulates other genes during development. Mutation of the MECP2 gene was identified as the cause of Rett syndrome by Dr. Huda Zoghbi and her laboratory in 1999. Since this seminal finding that links MeCP2 and RTT, several genetically modified mouse models of RTT have been generated. These mouse models were analyzed to determine whether MeCP2 protein levels are tightly regulated to MeCP2 function, and indeed, a loss of MeCP2 does yield similar features of RTT found in humans. Despite the advances in genetic studies of RTT, the mechanisms by which dysfunction of MeCP2 leads to neurological symptoms remain poorly understood, and thus the development of therapeutics is delayed. In this article, Dr. Zhou and his colleagues describe a new mouse model for RTT designed to study a human mutation (T158A) in MeCP2 that is associated with Rett syndrome.

[Read More...](#)

[Abstract](#)

[Article](#)

## Rett Syndrome and Sleep

A good night's sleep promotes learning, improved mood, general good health, and a better quality of life for both your child and the whole family. This article written for the International Rett Syndrome Foundation (IRSF) by Dr. Daniel Glaze, Medical Director of the Blue Bird Circle Rett Center and Sleep Disorder expert, reviews the benefits of sleep; the risks involved with disrupted sleep; the types of sleep problems experienced frequently in Rett syndrome; and strategies for getting back on track to good sleep habits. Many Frequently Asked Questions are answered!

[Read Full Article](#)

## Featured Angel: Katie Nues

Hi, my name is Katherine Nues but everyone calls me Katie. I am 9 ½ years old! I live at home in California with my sisters Melissa (5), Abby (4), and mom and dad. I love being with them, reading stories, going places, swimming together, listening to my favorite songs. I especially love when my dad plays guitar just for me, and helps me pluck the strings. I am in the 4th grade this year and have a full-time helper and lots of friends, both in my Special Ed home room and my Regular Ed classes. Science is my favorite. I love seeing my friends every day. They have fun helping me get around school in my wheelchair because it has light-up sparkle wheels :o) I like my chair too because I couldn't go anywhere without it. They also think my new computer is really cool, more about that in a minute...

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

## Fundraising and Hope Raising...

Take bold action this year and help us fight for our girls and women fighting Rett syndrome! Set your 2012 resolution NOW to get involved and take part. Every day our angels with Rett syndrome face the world with a brave face as they patiently wait for us to find treatments and a cure. IRSF has hit the ground running this year – make this the year you get active and give hope to thousands. Take part in a local IRSF Strollathon, host your own IRSF Signature Event, or create an online fundraising page! Click here for an A-Z list of easy fundraising ideas to get your office, kid's school or your community on board with fundraising for your Rett angel! IRSF makes it easy to get involved.

Your help is our hope. Your help is their hope.

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!

Host an Online Fundraiser:

As we all know, the holiday season is a very busy time! Instead of planning a fundraiser this winter why not to an online fundraiser! By using the FirstGiving online tool you can fundraise with just a few clicks of your mouse. Go to [www.firstgiving.com/irsfcareandcure](http://www.firstgiving.com/irsfcareandcure) to set up your very own fundraising page. Click here for instructions on how to set up your page. FirstGiving is easy to navigate and helps you personalize your page, post to your Facebook profile, and even walks you through how to email your page to your email contacts!

[Click here to download instructions.](#)

Join a Support Network

Find your Regional Rep



[Shop](#)

[Past Issues](#)

Natural History Study - Florida X

## Recent Publications

MeCP2 binds to nucleosome free (linker DNA) regions and to H3K9/H3K27 methylated nucleosomes in the brain.

Thambirajah AA, Ng MK, Frehlick LJ, Li A, Serpa JJ, Petrotchenko EV, Silva-Moreno B, Missiaen KK, Borchers CH, Adam Hall J, Mackie R, Lutz F, Gowen BE, Hendzel M, Georgel PT, Ausió J.

[Abstract](#)

[Article](#)

Anesthetic management of an adult patient with Rett syndrome and limited mouth opening –A case report

Nho JS, Shin DS, Moon JY, Yi JW, Kang JM, Lee BJ, Kim DO, Chung JY.

[Abstract](#)

[Article](#)

The diagnostic odyssey to Rett syndrome: The experience of an Australian family.

Knott M, Leonard H, Downs J.

[Abstract](#)

[Article](#)

Generation and Characterization of Rat and Mouse Monoclonal Antibodies Specific for MeCP2 and Their Use in X-Inactivation Studies.

Jost KL, Rottach A, Mildner M, Bertulat B, Becker A, Wolf P, Sandoval J, Petazzi P, Huertas D, Esteller M, Kremmer E, Leonhardt H, Cardoso MC.

Abstract

Article

Medical care of adolescents and women with Rett syndrome: An Italian study.

Vignoli A, La Briola F, Peron A, Turner K, Savini M, Cogliati F, Russo S, Canevini MP.

Abstract

Article

Normal mitral cell dendritic development in the setting of Mecp2 mutation.

Palmer AM, Degano AL, Park MJ, Ramamurthy S, Ronnett GV.

Abstract

Article

Methodological note: Video analysis of the early development of Rett syndrome-one method for many disciplines.

Marschik PB, Einspieler C.

Abstract

Article

FOXG1 mutations in Japanese patients with the congenital variant of Rett syndrome.

Takahashi S, Matsumoto N, Okayama A, Suzuki N, Araki A, Okajima K, Tanaka H, Miyamoto A.

Abstract

Article

Contributing to the early detection of Rett syndrome: The potential role of auditory Gestalt perception

Marschik PB, Einspieler C, Sigafoos J.

Abstract

Article

Subclinical myocardial dysfunction in Rett syndrome

De Felice C, Maffei S, Signorini C, Leoncini S, Lunghetti S, Valacchi G, D'Esposito M, Filosa S, Della Ragione F, Butera G, Favilli R, Ciccoli L, Hayek J.

Abstract

Article

Spinal Fusion for Scoliosis in Rett Syndrome With an Emphasis on Early Postoperative Complications.

Gabos PG, Inan M, Thacker M, Borkhu B.

[Abstract](#)

[Article](#)

Initial assessment of the StepWatch Activity Monitor™ to measure walking activity in Rett syndrome.

Downs J, Leonard H, Hill K.

[Abstract](#)

[Article](#)

Barriers to diagnosis of a rare neurological disorder in China-Lived experiences of Rett syndrome families.

Lim F, Downs J, Li J, Bao XH, Leonard H.

[Abstract](#)

[Article](#)

Expanding the phenotype associated with FOXP1 mutations and in vivo FoxG1 chromatin-binding dynamics

De Filippis R, Pancrazi L, Bjørge K, Rosseto A, Kleefstra T, Grillo E, Panighini A, Cardarelli F, Meloni I, Ariani F, Mencarelli M, Hayek J, Renieri A, Costa M, Mari F.

[Abstract](#)

[Article](#)

## Clinical characteristics of Rett Syndrome

Abbes Z, Bouden A, Halayem S, Othman S, Bechir Halayem M.

[Abstract](#)

[Article](#)

## Concomitant microduplications of MECP2 and ATRX in male patients with severe mental retardation

Honda S, Satomura S, Hayashi S, Imoto I, Nakagawa E, Goto YI, Inazawa J.

[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.

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

## 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](http://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](mailto:lhayden@rettsyndrome.org) or by fax at 513-874-2520

## Mail

Please send to 4600 Devitt Drive Cincinnati, OH 45246.

"Keep the main thing, the main thing."

~ Anonymous

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](http://www.rettsyndrome.org) or call IRSF at 1-800-818-7388 (RETT).

Designed by Songswift: global village solutions

© 2011 All Rights Reserved

