

IRSFlash: 3.9.2011

MARCH

9

## In This Issue

Research - IRSF Announce Planning for 7th World Congress

Happenings - Family Conference: Register NOW for Early-bird Discount

Upcoming Events - Find an Event Near You!

## Research

Investigator Spotlight: Lisa Monteggia, PhD, University of Texas Southwestern Medical Center

Dr. Lisa Monteggia of the University of Texas Southwestern Medical Center received a Regular Research Grant last year for her project titled "Elucidation of Epigenetic Mechanisms in Rett Syndrome." The project involves studying changes that occur in neurons as a result of mutations in MeCP2. This year Dr. Monteggia will Co-Chair a Session at IRSF's 12th Annual Rett Syndrome Symposium in Leesburg, VA.

Learn more about Dr. Monteggia and her research.

#### Genzyme takeover may boost interest in rare diseases

The field of rare diseases has gained more attention, not just from pharmaceutical companies but from the FDA and the NIH and is likely to draw more interest from other biotech companies, following the acquisition of the pharmaceutical company Genzyme, who are the world's largest maker of rare disease drugs. Genzyme previously formed a partnership with the company PTC therapeutics and have been working to develop the drug Ataluren which is currently being tested by researchers working on Rett syndrome.

For more on this story: [click here](#)

#### IRSF Announces Planning Underway for 7th World Congress on Rett Syndrome

Following the 6th World Congress on Rett Syndrome, which took place in Paris, France in 2008, IRSF agreed to host the 7th World Congress on Rett Syndrome and the Foundation is proud to say that planning has commenced. This 2012 international, interdisciplinary meeting will take place June 24th through June 28th, 2012. It will bring together speakers and luminaries in the field of Rett syndrome together with families, IRSF staff and a host of other colleagues and professionals in the culturally rich city of New Orleans, LA.

[Learn more](#)

Happenings

Featured Angel: Kaitlyn Kelly

My name is Kaitlyn Kelly and I am from Bladenboro, NC.

I am 4 years old and I am a Pre-K student in Mrs. Debbie Simmons class at Whiteville Primary School.

Mrs. Debbie is such a wonderful teacher; she actually is the one who told my mommy to have me checked out for Rett syndrome! (Thank you, Mrs. Debbie. More than you'll ever know).

Learn more about Kaitlyn and her family

Annual Family Conference 2011

Register NOW for Your Early Bird Discount!

"Cheers to Research to Reality"

May 27-30, 2011\*

The Boston Park Plaza Hotel

Boston, MA

Please join us at the IRSF Annual Family Conference this year. This conference is the only one of its kind where parents, caregivers, extended family and professionals come together to learn current care and advocacy strategies and the latest in research and clinical trials. \*Early Bird Rate: \$260 per person for Full Conference Registration and \$160 for Single Day Registration Before April 17th.

\*Early Bird Rate: \$260 per person for Full Conference Registration and \$160 for Single Day Registration Before April 17th.

We've packed our agenda with new and exciting speakers, exhibitors and more!

Motivational keynote by Massachusetts's own Dick Hoyt of Team Hoyt

Leading Research experts from Boston Children's, Harvard and MIT, including IGF-1 Clinical Trial Investigator Dr. Omar Khwaja

Communication experts Susan Norwell and Judy Lariviere

Sessions on therapies, education, family foundations, financial and legal life planning

First-ever sessions for the Spanish-speaker

First-ever entire track of sessions on the direction of research

Don't miss your chance to build and grow your knowledge, expertise and awareness of Rett syndrome. Plan ahead and take advantage of our early-bird registration prices. This is an investment in YOU!

[Register Now!](#)

### A Daughter's Legacy

Judy and Clifford Fry, of College Station, Texas, used this Rare Disease Day as a way to educate a new group of individuals about Rett syndrome. Just as we use October Awareness Month as our time to push advocacy for Rett syndrome, Judy and Clifford, parents of Ashley, spread awareness through an article in Insite Magazine. "The governor proclamation gave me the foot in the door with these folks," Clifford said. "I tried to get an article last year (October), but they put off publishing an article about Ashley until this February. Judy and I will still use this locally and with friends as well as in our fund raising efforts supporting the Texas Strollathons."

[Click here to read the article \(pp. 20-23\)](#)

You can also visit [www.rarediseaseday.org](http://www.rarediseaseday.org) to learn more about what others did on February 28th to spread awareness for the cause close to their hearts.

## Stakeholder Reviews

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!

## Brain Behavioral Relationships

Brain behavior relationships in Angelman Syndrome, Rett Syndrome, and MECP2 duplication syndrome.

We want to follow:

Children with Angelman syndrome, Rett syndrome, and MECP2 duplications.

Who are between the ages of 3-10.

For all participants:

We will do an assessment of your child's development and behavior.

Have your child look at images on a computer screen to track the movement of their eyes, and their responses to different facial expressions.

Have you complete forms about your child's behaviors and sensory interests/aversions.

You will receive a report of your child's developmental abilities at no cost.

We will compensate families for their time.

Contact: Amy.k.wilson@vanderbilt.edu (615) 322-8093

Be Bold. Get Active. Give Hope.

Take a BOLD action this year and help us fight for a cure for Rett syndrome! Set your 2011 resolution NOW to get involved and make a difference. Our Rett angels take on each day 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! IRSF makes it easy to get involved. Your help is our hope; your help is their hope.

Are you holding an event in 2011? Fill out this form and send back to Jenni Grammer at [jgrammer@rettsyndrome.org](mailto:jgrammer@rettsyndrome.org)

The Rett Gazette



Keep an eye out for the latest issue of the Rett Gazette which should hit mailboxes sometime between late March and early April! This issue is one of our best yet - packed with information on the Revised Diagnostic Criteria, Conference, research updates, recent news, family support updates and MORE! Don't miss your chance to educate yourself and stay connected.

Check out some of our past issues

Sign up to receive the Rett Gazette

Henhouse Coffee Supports Rett Syndrome & You Can HELP!

IRSF is excited to announce a new fundraising partnership with Henhouse Coffee. Henhouse Coffee is a Colorado based specialty coffee roasting company. 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) to place your order for Henhouse Coffee and to lend support to IRSF. Browse the website to learn more about Henhouse, the art of coffee roasting and discover which brew is right for you. When you've decided, 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!

Try out our NEW 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.

## A Glance @ Rett

[Join a Support Network](#)

[Find your Regional Rep](#)

[Shop](#)

Friend us on Facebook

Follow us on Twitter

## Past Issues

## Recent scientific articles related to Rett Syndrome

Isolation of MECP2-null Rett Syndrome patient hiPS cells and isogenic controls through X-chromosome inactivation

Authors: Cheung AY, Horvath LM, Grafodatskaya D, Pasceri P, Weksberg R, Hotta A, Carrel L, Ellis J.

Journal Citation: Hum Mol Genet. Mar 3, 2011

Characteristics of late-onset epilepsy and EEG findings in children with autism spectrum disorders

Authors: Lee H, Kang HC, Kim SW, Kim YK, Chung HJ.

Journal Citation: Korean J Pediatr. 2011 Jan;54(1):22-8 – Free Article

#### Upcoming Events

3rd Annual Boston Bruins Alumni Game

Natural History Research Study - Oakland

Natural History Research Study - Chicago

Riverside Rett Lunch And Auction

Rett Spring Fling

Annual Family Conference

Memorial Day Weekend, May 27-30, 2011\*

The Boston Park Plaza Hotel

Boston, MA

12th Annual Rett Syndrome Research Symposium

June 26 – 28, 2011

The Lansdowne Resort and Spa, Leesburg, Virginia.

Pharmacological Treatment of Rett Syndrome with IGF-1

After Phase 1 determines the safety of IGF-1 therapy for girls with Rett syndrome, phase two will conduct a 20 weeks long treatment program in which the girls are seen monthly.

Rett Syndrome Research Moves Even Closer to Reality

IRSF approved additional Translational Research grant awards in December, bringing 2010 to a close with a record \$2.7 million in funded research grants out of a total direct research spend of \$3 million.

Rett Syndrome Natural History Study

We have several enrollment openings for the Chicago spring data collection April 9-10, 2011.

Rett Care Survey

This survey is being conducted by Rett parent, Henry Perez (daughter Lily, age 14), who is working on his dissertation. His research is based on home and community based services and the integration of individuals with Intellectual and Developmental Disabilities (IDD).

[Click here to take the survey](#)

\*Note: This survey will be available through April 15th

Vote for a Sponsafier Rett Syndrome Car!

Car 11: Cure Rett Syndrome

Car 55: Rett's Awareness

## In the News

[Restrictive licensing delays research into rare disease](#)

[Shelby coach steps down to help son battle autism](#)

[Repairing faulty genes](#)

[FAQ about the IGF-1 Rett Syndrome Trial](#)

[MeCP2: Structure and Function](#)

[When my son smiles...](#)

[Age-dependent expression of MeCP2 in a heterozygous mosaic mouse model](#)

[Rett syndrome – MECP2 mutations, faulty instructions for making protein \(MeCP2\) critical for normal brain development and forming synapses](#)

[February's Mom of the Month \(Hint: It's a Rett syndrome parent\)](#)



## 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.

## United Way

While we don't have a UW code, many will accept donor directed donations. Write our EIN - 31-1682518, name & address on the donor designation line.

## 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 cleighton or by fax at 513-874-2520

## Mail

Please send to 4600 Devitt Drive Cincinnati, OH 45246.

## Rett Quote

"Life's greatest rewards often appear in unexpected places."

~ Unknown

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 media solutions

© 2011 All Rights Reserved