

IRSFlash: 6.23.2011

June

23

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A Message from IRSF Executive Director, Stephen Bajardi

We've just returned from an incredible annual family conference in Boston over the Memorial Day weekend. Over 300 people convened for an outstanding three day offering of sessions on family support and research. There were many highlights, but of particular interest was an announcement by IRSF Chair Kathryn Kissam of a recent gift from the Pioneer Fund of Colorado to partially fund a new, national public awareness campaign featuring Clint Black. This campaign will blanket the country during October Rett Syndrome Awareness Month; it will be distributed to 210 television and radio markets throughout the United States and spearhead our efforts to find new families and friends. The research presentations were not only incredible, they were very encouraging; the highlight was Dr. Omar Khwaja who reported some positive results on the Rett syndrome IGF1 trial he directs in Boston; his report had everyone on the edge of their seats. Participants at the meeting were treated to a tour of his lab, those who attended were amazed at the work of his team. There were many sessions on family support; among the most memorable and exciting was one by Dr. Pam Diener of Marymount University. Dr. Diener reported on a Hippo therapy study and had data to show positive improvement on mobility and general health. Prior to the patient conference there was an IRSF Board meeting where new Board member Bill Farnum was thanked for his outstanding work with Nike and voted onto the national Board. IRSF is in a strong position, coming off a great year to push the Research to Reality agenda of accelerating research to develop treatments and a cure. The plan for Research to Reality is moving forward and this fall it will move to new levels of fundraising, spreading awareness, advocacy and science. The high energy and feeling of closeness to each other dominated the meeting and among the most memorable experiences were the awards ceremony Saturday night and a performance by Amy (Sanchez) Loren, the folksinger who created the song "Silent Angels" for Rett syndrome. It was a wonderful meeting, a good way to bring the holiday weekend in.

I am sure you are all working on care issues for your children for the summer. With school out and vacations planned, establishing care for the children is the primary concern. I hope everyone takes the time to renew your energy, experience high quality family time and get ready for a very ambitious fall. We have letters to write and visits to make with our Congressman and Senators to make sure Rett syndrome is not lost in the budget discussions. There will be TV and radio stations to visit, so people can see and get to know Rett syndrome in ways they haven't before. There are

huge opportunities in science that need sufficient funding if we are to see the results we all want and families will need support every day. I wish everyone a great summer. We will be watching closely, on your behalf, events in DC and elsewhere. We will be busy preparing for the big awareness effort this fall and we are looking forward to seeing everyone participate in the most active October Rett Syndrome Awareness Month yet.

Rest up! Have a good summer, see you soon!

Stephen Bajardi

IRSF's 12th Annual Rett Syndrome Symposium to take place Sunday, June 26th – Tuesday, June 28th 2011 in Leesburg, Virginia is at full capacity!

We're excited to report that we have experienced a record interest in attending this year's meeting--especially from the international community.

Next year IRSF will host the 7th World Congress on Rett Syndrome to take place June 24th - June 28th, 2012 in New Orleans, Louisiana. This very important, global meeting 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. Please lookout for further communication on this meeting as the details become available.

[Read more...](#)

IRSF Welcomes New Board Member: Bill Farnum

Bill Farnum of Nike Inc., an active parent in the Rett syndrome community, has been elected to the IRSF Board of Directors. Bill lives in Oregon with his wife, Beth, and their daughter, Ella, who has Rett syndrome. "The board is eager to work with such a dedicated and energetic individual to create a world without Rett syndrome," said chair of the board, Kathryn Kissam. Over the last three years, Bill's annual event, Casting 4 A Cure, has grown into a multi-faceted, non-profit organization that raises critical funds for research and support for families affected by Rett syndrome. Since its inception, Casting 4 A Cure has raised over \$125,000 for Rett syndrome research and family support.

This year's fly fishing tournament will be held July 21-23 in Steamboat, CO and August 25-28 in Victor, ID.

Research

Investigator Spotlight: Yi Eve Sun, PhD, University of California, Los Angeles

With this month's installment my goal to expose as many IRSF funded investigators who have a primary role in IRSF's 12th Annual Rett Syndrome Symposium to take place June 26th through 28th at the Lansdowne Resort and Spa in Leesburg, Virginia comes to a close. This month I saw fit to profile one of our meeting Chairs, Dr. Yi Eve Sun from the University of California, Los Angeles who is also a Co-Chair of this year's Session on animal and human cellular models of Rett syndrome (RTT) and is also a 2010 IRSF HeART Grant Award recipient. Her project titled "A high throughput small molecule screening platform for potential Rett Syndrome MBD mutation therapeutics" involves a search for candidate drugs that may enhance the binding of mutant MeCP2 protein to 'methylated' DNA. This work is important because RTT mutations frequently arise in the methyl-CpG binding domain (MBD) of the MECP2 gene. Such mutations of the gene are thought to result in the production of a misshapen MeCP2 protein that can no longer bind to methylated DNA. Dr. Sun's drug screen will be conducted using both biochemical test-tube based screens, in combination with cell-culture based screens (using stem cells with common RTT mutations). Taking this approach she hopes, may lead to the discovery of new drugs specifically targeted for individuals with RTT caused by MBD mutations.

Learn more about Yi Eve Sun, PhD.

Focus of Seizures is Identified in Mecp2 Mouse

Focus of Seizures is Identified in Mecp2 Mouse

A region of the brain called the hippocampus, is one of the main areas responsible for the genesis epilepsy in the brain. Due to its high degree of inter-connectivity, the proper development of neurons in the hippocampus leads to a finely tuned "excitation-to-inhibition balance". Keeping this balance is absolutely critical, however, it has been recently found that this excitation-to-inhibition balance is dysfunctional in several mouse models of autism spectrum disorders. Recent work by Dr. Lucas Pozzo-Miller and team describes for the first time a pronounced network hyper-excitability that is caused by an excitation-to-inhibition imbalance in the hippocampus of Mecp2 mutant mice.

- Network hyperexcitability in hippocampal slices from Mecp2 mutant mice revealed by voltage-sensitive dye imaging 60.34 Kb

Developmental Disease is Re-created in an Adult Model

We are delighted to share with you this press release which outlines an IRSF funded study showing that Rett syndrome can be reestablished in an adult animal model. IRSF funded research continues to make advancements in our search for treatments and a cure for Rett syndrome. Kathryn Kissam, Chairman of the IRSF Board, remarked, "We are gratified that the research produced by Dr. Zoghbi's lab continues to show great advances. Coming off of the research reported at the Annual Conference, this is indeed additional good news for Rett syndrome."

- Read The Press Release
- Huda Y. Zoghbi, MD, Receives \$500,000 Gruber Neuroscience Prize

Happenings

Help IRSF Win up to \$250,000 from Vivint

The International Rett Syndrome Foundation is competing in the [vivint.givesback PROJECT](#). Cast your DAILY Facebook vote for IRSF EVERY DAY from June 14th through AUGUST 27th. You can vote for ONE CHARITY PER DAY! The charity that earns the most overall votes will then be awarded \$250,000. The remaining charities that earn the most from each of the regions will each receive a \$100,000 donation! Vote and share EVERYDAY!

Sign up for DAILY EMAIL REMINDERS

Want to Know More...

Can I vote if I am not a U.S. citizen?

Yes. Everyone with a Facebook log in can participate in the voting process.

Can I vote if I'm not on Facebook?

Unfortunately, this voting competition is run solely on Facebook. We are aware that not everyone utilizes Facebook, but hope that you might still support us by spreading the word via email, phone, etc. to all of your friends and family who do use Facebook and ask them to vote daily. You can still be an key part of this competition just by vote-raising for IRSF.

Cheers to Annual Family Conference 2011!

Over 300 people joined IRSF over Memorial Day weekend to meet and learn from top Rett syndrome experts at our 27th annual Education and Awareness conference in Boston, MA. Of special interest to attendees this year was the first-ever dedicated session track on research, led by leading Rett syndrome researchers from Boston Children's Hospital, Harvard, MIT, Baylor College of Medicine, the University of Alabama Birmingham, Georgetown University, University of Western Ontario and more. Especially well received were the early results reported by Omar Khwaja, MD, PhD, director of the Rett Syndrome Program in the Department of Neurology at Children's Hospital Boston, on his newly begun IGF-1 clinical trial. The conference began on Friday afternoon with a private Children's Hospital Boston lab tour, where many parents for the first time ever saw a full 360 degree bench-to-bedside approach to Rett syndrome Basic to Translational and Clinical Studies in action.

On-demand internet replay of the 13 Saturday sessions now available for purchase for only \$100 (additional 25% discount for paid conference attendees – call 1.800.818.RETT (7388) to order)

Throughout the rest of the weekend, families and professionals chose from over 40 unique sessions focused on Research, Communication and School, Medical Issues and Therapies, Family Foundations, Fundraising and Advocacy. Also joining us were over 15 interactive Exhibitors, demonstrating products and services of specific interest and benefit to

our children living with Rett syndrome. The very inspirational keynote motivational talk delivered by Massachusetts's own world-renowned triathlete and marathoner Dick Hoyt of Team Hoyt, sponsored by the Rett Syndrome Association of Massachusetts, left all attendees with the bold takeaway message "YES YOU CAN"!

Attendees also enjoyed refreshing Pepsi products throughout the weekend thanks to a generous donation from the Pepsi Beverages Company - New England and Tri-State MU in recognition of IRSF's \$250,000 win in the 2010 Pepsi Refresh Project.

The Tribute Reception was an incredibly special highlight event hosted in the grand historic Imperial Ballroom at the Boston Park Plaza Hotel. The program opened with a unique and lovely Boston cultural Irish Step Dance performance, followed by the annual award ceremony, viewing of the 2011 Tribute video, and an amazing live concert performance featuring Ayme (Sanchez) Loren, singing her hit song "Silent Angels" with guitarist Eduardo Toledo Guimaraes.

2011 AWARD RECIPIENTS

Art of Caring Award recipient Dr Omar Khwaja, Director Rett Syndrome Program, Children's Hospital Boston

Each year at the Tribute Reception, IRSF recognizes individuals who have made significant contributions towards IRSF's mission to advance research for treatments and a cure for Rett syndrome while improving the overall quality of life for those living with Rett syndrome today. The following awards were presented by IRSF's Board of Directors to:

- Omar Khwaja MD, PhD, Director Rett Syndrome Program, Children's Hospital Boston - Art of Caring Award, IRSF's most prestigious award for exceptional devotion to Rett syndrome
- Jeffrey Neul, MD, PhD, Assistant Director, Blue Bird Circle Rett Center, Baylor College of Medicine - Circle of Angels Award, for his enduring commitment to Rett syndrome biomedical and clinical research
- Donna Wright, grandmother to Naomi - Giving Hope a Hand Award, for her inspiration acts of advocacy and awareness
- Henry Perez, father to Lily - Volunteer of the Year Award, for his overall outstanding dedication to IRSF's mission

IRSF congratulates and thanks our award recipients for going above and beyond every day for our children.

We appreciate all attendees, planning committee members, speakers, exhibitors, volunteers, vendors and staff for making this conference a tremendous success. A special thanks goes to our conference collaborators at Boston Children's Hospital and the Rett Syndrome Association of Massachusetts!

Please visit www.rettsyndrome.org/conference for full Conference Program information. Event photos, session slides, handouts, and information on orderable session DVD s and audio MP3s will be made available shortly.

Featured Angel: Cameron Babiarz

Hi, my name is Cameron, and I was born on March 10, 2009. My parents had concerns when I was six months old and did not show interest in transitioning or crawling. Doctors said "wait and see" for the next six months. When I was 12 months old, testing continued, and I began physical, occupational, developmental, and speech therapy. In the meanwhile, my MRI and blood work all came back normal. By 18 months, I was still unable to sit, transition, crawl, pull to stand, or walk. I lost my verbal communication and self-feeding and manipulations skills, and my left hand was constantly in my mouth. At 20 months, we visited a physiatrist who made a clinical diagnosis of Rett syndrome. On January 7, 2011 at 2:45 pm, my mommy, who was 35 weeks pregnant, received a call confirming that I had Rett.

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

Congratulations Vera!

IRSF would like to congratulate Rett supporter and Tennis star, Vera Zvonareva, who was seeded second, beat Alison Riske of the United States Monday in the first round at Wimbledon.

Vera, an avid Rett syndrome supporter, attended a special Meet and Greet with a few local girls with Rett syndrome last summer during her time here while competing in the Western Southern Financial Group Masters and Women's Open.

Zvonareva serves as an international ambassador for Rett syndrome and spreads awareness around the world when she travels to tournaments. She became familiar with Rett syndrome after her best friend's daughter was diagnosed.

IRSF would like to express our unwavering support for Vera at Wimbledon! Good luck Vera and thank you!

- [Read More](#)
- [Vera and Rett syndrome](#)
- [Make a Facebook post on the IRSF Facebook page or on Vera's Facebook page!](#)

Fundraising and Hope Raising...

Over the years, IRSF volunteers, parents, caregivers and advocates have used fundraising to do more than raise crucial research funding – it has helped instill hope in the community, and has helped those with loved ones suffering from Rett syndrome to heal. As a volunteer-driven Foundation, IRSF prides itself in supplying their dedicated volunteers with the skills, materials and support they need to make their fundraising successful.

Over the last 10 years the momentum being built in the community is unmatched. The evolution of the IRSF fundraising program is proof that hope does drive many and getting involved is contagious. **THIS IS THE YEAR TO TAKE ACTION!** Many of you already have planned fundraising events – are they listed on the IRSF calendar? If not, please fill out this form and email to Jenni Grammer at jgrammer@rettsyndrome.org. Also check the website calendar for events in your neck of the woods! IRSF events are growing and growing. Do you have an idea for an event and need help getting started? That's what the IRSF Staff is here for! Reach out to us, let us help you. Contact the IRSF office at 1800-818-7388 (RETT) to ask questions, get information and answers or if you just need help brainstorming.

Your help is our hope. Get involved!

One More Challenge

One More Challenge is the heartwarming story of a family with 16 children-2 of them being severely handicapped. Told from a father's point of view, the day to day ins and outs, the dramatic accomplishments and tragedies that are present in the lives of these "ordinary people" will surely catch your interest and warm your heart!

[Click here](#) to get more information on how you can get your very own copy of this heart warming story.

[A Glance @ Rett](#)

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Past Issues

Tell a Friend

Upcoming Events

Natural History Research Study - Florida

6th Annual Cape Cod Strollathon

12th Annual Scientific Symposium

1st Annual Cure For Keira Golf Tournament

3rd Annual Cierney's Ride For Rett

9th Annual Rett Syndrome Road Tour

Casting 4 A Cure Fly-Steamboat, CO

Annual OH/KY/IN Family Picnic

8th Annual IRSF Charity Golf Classic

Recent scientific articles related to Rett Syndrome

Recent Publications

Ex Vivo Treatment with a Novel Synthetic Aminoglycoside NB54 in Primary Fibroblasts from Rett Syndrome Patients Suppresses MECP2 Nonsense Mutations

Vecsler M, Ben Zeev B, Nudelman I, Anikster Y, Simon AJ, Amariglio N, Rechavi G, Baasov T, Gak E,

PLoS ONE 6(6), June 13, 2011

Sex differences in epigenetic mechanisms may underlie risk and resilience for mental health disorders

Jessen HM, Auger AP.

Epigenetics. 2011 Jul 1;6(7).

Investigation of modifier genes within copy number variations in Rett syndrome

Artuso R, Papa FT, Grillo E, Mucciolo M, Yasui DH, Dunaway KW, Disciglio V, Mencarelli MA, Pollazzon M, Zappella M, Hayek G, Mari F, Renieri A, Lasalle JM, Ariani F. J Hum Genet. 2011 May 19.

Cognitive deficits in Rett syndrome: What we know and what we need to know to treat them

Berger-Sweeney

J. Neurobiol Learn Mem. 2011 May 23.

Oligogenic heterozygosity in individuals with high-functioning autism spectrum disorder

Schaaf CP, Sabo A, Sakai Y, Crosby J, Muzny D, Hawes A, Lewis L, Akbar H, Varghese R, Boerwinkle E, Gibbs RA, Zoghbi HY.

Hum Mol Genet. 2011 May 30.

Biogenic amines and their metabolites are differentially affected in the Mecp2-deficient mouse brain

Panayotis N, Ghata A, Villard L, Roux JC. BMC Neurosci.

2011 May 24;12(1):47.

Effects of Creatine Supplementation in Rett Syndrome: A Randomized, Placebo-Controlled Trial

Freilinger M, Dunkler D, Lanator I, Item CB, Mühl A, Fowler B, Bodamer OA.

F(4)-neuroprostanes mediate neurological severity in Rett syndrome

Signorini C, De Felice C, Leoncini S, Giardini A, D'Esposito M, Filosa S, Della Ragione F, Rossi M, Pecorelli A, Valacchi G, Ciccoli L, Hayek J.

Clin Chim Acta. 2011 Apr 17.

Past Publications

Adult Neural Function Requires MeCP2

McGraw CM, Samaco RC, Zoghbi HY. Science. 2011 Jun 2

Clin Chim Acta. 2011 Apr 17.

Complexities of Rett Syndrome and MeCP2

Motil KJ, Barrish JO, Lane J, Geerts SP, Annese F, McNair L, Percy AK, Skinner SA, Neul JL, Glaze DG.

J Pediatr Gastroenterol Nutr. 2011 May 30.

Vitamin D Deficiency is Prevalent in Females with Rett Syndrome

Foley KR, Downs J, Bebbington A, Jacoby P, Girdler S, Kaufmann WE, Leonard H. J Child Neurol. 2011 Jun 2

Change in Gross Motor Abilities of Girls and Women With Rett Syndrome Over a 3- to 4-Year Period

Signorini C, De Felice C, Leoncini S, Giardini A, D'Esposito M, Filosa S, Della Ragione F, Rossi M, Pecorelli A, Valacchi G, Ciccoli L, Hayek J.

Clin Chim Acta. 2011 Apr 17.

Epilepsy Treatment in Rett Syndrome

Krajnc N, Zupancic N, Orazem J.

J Child Neurol. 2011 Jun 2

Habituation without NMDA Receptor-Dependent Desensitization of Hering-Breuer Apnea Reflex in a Mecp2 Mutant Mouse Model of Rett Syndrome

Song G, Tin C, Giacometti E, Poon CS.

Front Integr Neurosci. 2011;5:6.

Rett syndrome: A study of the face

Allanson JE, Hennekam RC, Moog U, Smeets EE.

Am J Med Genet A. 2011 May 27

How epigenetics meet neurobiology in Rett syndrome: topics in the 2010 Kavli Prize Symposium on Neuroscience

Bergersen LH, Sander M, Storm-Mathisen J.

Neuroscience. 2011 May 27.

In the News

Partnership to host 'Disability Awareness Day' at Patriots Stadium

Role of MeCP2 in Regulating Synapse Function and Behavior

Role of MeCP2 (Methyl CpG-Binding Protein) during replication

Atlantic Health Hospitals - Treating Rett Syndrome

Rett Syndrome - Nursing Link

Support Rett Syndrome

NEW: 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. This page will be primarily run by you, so feel free to post and comment as you'd like. IRSF will moderate as needed; however YOU decide how to best use this page for your state! Search for your state's page by "(State) IRSF Rett Syndrome Families." You may have to request to join the page before you can start posting!

Find out if your state has a page! Here are a few:

Alaska IRSF Rett Syndrome Families

California IRSF Rett Syndrome Families

Connecticut IRSF Rett Syndrome Families

Florida IRSF Rett Syndrome Families

Iowa IRSF Rett Syndrome Families

Nevada IRSF Rett Syndrome Families

New York IRSF Rett Syndrome Families

North Carolina IRSF Rett Syndrome Families

Pennsylvania IRSF Rett Syndrome Families

Rhode Island IRSF Rett Syndrome Families

Virginia/DC IRSF Rett Syndrome Families

Washington IRSF Rett Syndrome Families

Morgan's Wonderland

The first amusement park in the USA, and I suppose the world, designed for the handicapped. [Read More...](#)

The Rett Gazette

Don't receive the Rett Gazette?

Check it out online!

Sign up to receive the Rett Gazette

Autism Speaks Announces New Science Funding

Autism Speaks is pleased to announce its first research grants of 2011 totaling more than \$1.4 million. One of the projects, the result of a Suzanne and Bob Wright Trailblazer Award, is a cutting edge study on gastrointestinal issues, which affect many children and adolescents with autism. [Read more about the other funded studies here.](#)

-e-Speaks, April 15, 2011

We NEED YOUR 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!

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 Foundation

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 "EIN - 31-1682518, IRSF, 4600 Devitt Dr, Cincinnati, OH 45246" 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 lhayden@rettsyndrome.org or by fax at 513-874-2520

Mail

Please send to 4600 Devitt Drive Cincinnati, OH 45246.

Rett Quote

"Values...know them, believe them, live them."

~ 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.rett syndrome.org or call IRSF at 1-800-818-7388 (RETT).

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