

IRSFlash: 1.10.2011

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To the Members of the Rett Syndrome Community:

On behalf of our Chair, Kathryn Kissam, the National Board, the Family Advisory Board, Regional Representatives, Research to Reality Chair, Clint Black, the Volunteer Scientific Leadership and National Staff, I send you all best wishes

for the New Year and extend a warm and grateful thank you for supporting IRSF this past year.

I especially want to thank all Strollathon, gala and golf tournament chairs and their event committees for their hard work as well as recognize the financial support from family led events of all kinds, donations from annual donors, exceptional gifts from individuals, the Pioneer Fund, the Pepsi Challenge, MSNBC, the Albemarle Foundation and the selfless work of thousands of volunteers including Congressman Steny Hoyer, his staff and Governors in almost 20 states.

IRSF raised over \$5.5 million in 2010, an incredible accomplishment and our best year ever! IRSF spent over \$3 million on direct research, the most ever in one year, held the largest family conference, distributed more useful information and provided more direct support to families than ever.

The New Year will be even more ambitious; the IRSF leadership has embraced a three year Strategic Research Plan which will push the boundaries of basic research and successfully build our translational research program. We plan to bring two, possibly more, compounds to eventual new clinical trials over the next three years.

The entire IRSF board and staff is committed to working for you to see the three year Strategic Research Plan successfully implemented and, with your support, we will succeed; working together we will take Research to Reality!

Your help is our hope; thank you for all you do.

Sincerely yours,

Stephen Bajardi

Executive Director

Investigator Spotlight: Dr. Omar Khwaja

In 2009 IRSF agreed to fund a Research Clinical trial conducted by Dr. Omar Khwaja and his team at Children's Hospital Boston. The proposal titled "Pharmacological Treatment of Rett Syndrome by Stimulation of Synaptic Maturation with IGF-1," represents the first potential disease-modifying therapy to be tested in RTT patients. This was greeted with an outpouring of enthusiasm from the Rett syndrome community.

[Learn more about Dr. Khwaja and his research](#)

Pharmacological Treatment of Rett Syndrome by Stimulation of Synaptic Maturation with IGF-1

This trial consists of two phases. Phase 1 is an intensive 6-week pharmacokinetic study which will require 3 inpatient stays and 4 half-day outpatient visits. The primary goal of Phase 1 is to determine the safety of IGF-1 therapy for girls with Rett syndrome. Phase two consists of an additional 20 weeks of treatment with IGF-1 where girls are seen monthly for safety monitoring, developmental evaluations and lab work. During this phase the research team will also monitor heart function (EKG) and electrical activity in the brain (EEG) four times.

[For more information on this research clinical trial or to enroll, please view our website.](#)

Rett Syndrome Natural History Study

The purpose of the study is to examine the progression of clinical features, quality of life, and longevity over a period of 10 years. We will gain broader insights into the many problems and idiosyncratic features associated with Rett syndrome.

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

[For more information on this research clinical trial or to enroll, please view our website.](#)

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.

The Advanced Neurotherapeutic Grant of Excellence (ANGEL) award mechanism provides large grant awards to target programs that help to identify new drug targets and to develop and test novel therapies for RTT. The second ANGEL award approved for funding in 2010 was provided in support of the following study:

Monica J. Justice, PhD from Baylor College of Medicine, titled “Developing new therapeutic targets for amelioration of Rett Syndrome from the identification of genetic suppressors in mice”

IRSF’s HELP ACCELERATE RETT THERAPEUTICS (HEART) grant awards provide seed funding to foster innovative projects at early stages of the drug discovery and development. Additional HEART grants awarded by IRSF in December 2010 were provided in support of the following studies:

Megumi Adachi, PhD from the University of Texas Southwestern Medical Center at Dallas, titled “Evaluation of Antidepressants as Potential Therapeutic Intervention for Rett Syndrome”

Kerry Delaney, PhD from the University of Victoria, titled “MeCP2 Replacement Using ‘Trojan Horse’ Pegylated Immunoliposomes”

Aleksandra Djukic, MD, PhD from Montefiore Medical Center, Einstein College of Medicine, titled “Cognitive Abilities in Patients with Rett Syndrome (Visual attention and Recognition)”

Stavros Lomvardas, PhD from the University of California, San Francisco, titled “A high throughput screen for chemicals that reverse the molecular deficits of MeCP2 deletion”

David J. Segal, PhD from the University of California, Davis, titled "Epigenetic Therapy for Rett Syndrome"

Xinyu Zhao, PhD from the University of New Mexico, School of Medicine, titled "Novel method for enhancing BDNF protein expression"

[Click Here](#) for additional information on research grants awarded in 2010.

IRSF announces the availability of grants in conjunction with both its Basic and Translational Research programs for 2011
Basic Research Program (Regular Research Grants & Post-doctoral Fellowship Awards)

Translational Research Program (HeART and ANGEL Grant Awards)

For individual program details, please visit our website.

Save the Date - 12th Annual Rett Syndrome Research Symposium

The International Rett Syndrome Foundation (IRSF) will host the 12th Annual Rett Syndrome Symposium from June 26th through June 28th 2011 at the Lansdowne Resort and Spa, Leesburg, Virginia. This year the symposium will be chaired by Yi Eve Sun, Ph.D. (University of California – Los Angeles) and James H. Eubanks, Ph.D. (University Health Network, Toronto Western Research Institute).

Please stay tuned for the full list of participants including our keynote speaker and session Chairs. [Click Here](#) to view the full agenda for last year's meeting.

NIMH's Top 10 Research Advances of 2010

In a recent post by the Director of the National Institute of Mental Health (NIMH) 10 research breakthroughs made in 2010 are changing the way we approach the development of treatments for nervous system disorders like Rett syndrome. Dr. Thomas Insel, suggests that one of the most important discoveries in the past decade was the creation of induced pluripotent stem cells (iPSCs): cells taken from adults, which have the potential of becoming any cell type. As a prime example, this revolutionary technology was recently applied to develop mature nerve cells using skin cells taken from an adult with Rett syndrome. The study by Marchetto and colleagues was recently published in the leading scientific journal *Cell* and was part funded by an IRSF grant to one of the investigators who participated in the study.

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

Funding Opportunity Announcements Related to Intellectual and Developmental Disabilities

The National Institutes of Health (NIH) have released several new requests for applications

[Click Here](#) for application details.

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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. We need you now more than ever!

Featured Angel: Haylee Bautista

“Aloha! My Name is Haylee Bautista! I am 5 years old. I am Pre-K and attend Kahului Elementary School in Maui. I also receive physical, occupational and speech therapy from Kahului Elementary.”

[Learn more about Haylee and her family](#)

Be sure to keep an eye out for Haylee’s featured day on the IRSF Facebook page!

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

Educators Network

Attention All Parents! IRSF has made it easy for you to get your daughter's teachers, therapists and all Rett-related educators in her life registered to become part of the IRSF Educators Network! Simply download, personalize and share this letter with anyone you feel would benefit from IRSF's new Educators Network.

Join the Educators Network

[Click here to download our Educator Invitation Letter \(for parents & Caregivers\)!](#)

REMINDER: Research Study Recruiting Parents of Children with Rett Syndrome

If you are the parent of a child with Rett syndrome, then there is a research study regarding your child's diagnosis that is seeking your participation!

A very big thank you goes out to all of the parents who have completed this survey already! If you have not yet taken this survey, or you know a fellow parent who has not taken it, now is your chance. The survey will be up through January 24th, 2011. I encourage all parents to participate, whether they have had their child tested or not. The more experiences I hear about, the better I can understand the impact genetic testing for Rett syndrome has had on families.

~ Christine Delaney, graduate student and researcher in genetic counseling at Brandeis University

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Upcoming Events

Find an Event Near You!

Educate Yourself

Annual Family Conference

Memorial Day Weekend, May 27-30, 2011*

The Boston Park Plaza Hotel

Boston, MA

www.bostonparkplaza.com

Travelers with Disabilities and Medical Conditions

One of the primary goals of the Transportation Security Administration (TSA) is to provide the highest level of security and customer service to all who pass through our screening checkpoints... TSA's checkpoint security screening procedures for persons with disabilities and medical conditions have not changed as a result of the current threat situation. All disability-related equipment, aids, and devices continue to be allowed through security checkpoints once cleared through screening.

[Read more](#)

Silent Angels

By Ayme Sanchez

Download the new single, Silent Angels, by Rett supporter and singer/songwriter Ayme Sanchez. Ayme co-wrote the song Silent Angels in honor of our Rett angels.

Download the song now from the IRSF iFan store on Facebook.

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.

"Make excellence an everyday event."

~ Character from Everyday Excellence