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Research

Investigator Spotlight: Zhaolan “Joe” Zhou, PhD, University of Pennsylvania, School of Medicine

Dr. Zhaolan “Joe” Zhou of the University of Pennsylvania, School of Medicine is a 2010 IRSF Regular Research Grant recipient. His project titled "The study of Rett syndrome with Mecp2 T158A knockin mice" involves the creation of a Rett syndrome mouse model that recapitulates a Rett mutation. This new mouse model should provide a closely relevant platform to develop and assess therapeutic treatments for Rett syndrome. Dr. Zhou will Co-Chair this year's Session on the regulation and function of MeCP2 at IRSF's 12th Annual Rett Syndrome Symposium in Leesburg, VA.

Learn more about Dr. Zhou and his research.

IRSF Funded Researchers Develop a "Green" Rett Mouse

The majority of individuals with Rett syndrome are female and are considered genetically ‘mosaic’ due to random inactivation of the X-chromosome (XCI) which occurs in around half of all their cells. As a result, for a girl with Rett syndrome, roughly half of all her cells will express genes from the chromosome containing the mutation in MECP2, randomly throughout her entire body. In order to better understand this phenomenon, a team of investigators at the University of New Mexico developed a novel “Green” Rett mouse. The team, led by IRSF funded researcher Dr. Xinyu Zhao crossed two existing genetic mouse models - one in which half of all cells in the animal lack a functional copy of mecp2 while the other model has a normal copy of mecp2, but also co-expresses green fluorescent protein (GFP) – a protein first isolated from jellyfish that is commonly used as a way of labeling cells. Now researchers for the first time have a way of looking at cells in a mouse brain to find out which are expressing a normal versus a mutant copy of the MeCP2 gene. Given the important function of MeCP2 in neuronal development, data emerging from the new mouse model sheds light on how MeCP2 loss affects postnatal brain function and highlights dynamic changes in the neurological symptoms of RTT later in development.

The study was carried out by researchers based at the University of New Mexico and published in an advance online in the journal Human Molecular Genetics.

[Link to Article](#)

Newly discovered protein could improve understanding of epilepsy

Epileptic seizures are a common complication of Rett syndrome and knowing more about how the signals in the brain work helps scientists better understand why, and when neurological disorders like epilepsy strike. The brain works like an electrical circuit, sending impulses along nerve fibers in the same way that current is sent through wires - when these

impulses are disrupted disorders such as epilepsy can occur. A team of investigators led by Prof. Peter Brophy at the University of Edinburgh in the U.K. have discovered that a "maintenance" protein which helps keep nerve fibers in the brain operating smoothly plays a key role in maintaining nerve axons – the parts of a nerve that transmit electrical impulses. While these findings may only be indirectly related to the seizures that occur in Rett syndrome, the study could point to new avenues to investigate for ways to prevent seizures and motor dysfunction.

The study was published in a recent issue of the Journal Neuron.

[Click here for more.](#)

Elite award for Adrian Bird

Adrian Bird, the University's Buchanan Professor of Genetics, has received one of the world's most esteemed medical prizes.

The scientist has been selected for a Canada Gairdner International Award. He received the honour for "pioneering discoveries on DNA methylation and its role in gene expression."

[Read More...](#)

Oral Calcium Supplementation to Treat Osteopenia in Rett Syndrome

What do we know?

Dietary factors may play an important role in bone health in Rett syndrome (RTT) and may be helpful in treating osteopenia (bone mineral loss), and bone fractures which frequently occur in girls and women with RTT. From an initial study we carried out, in which nearly 1,000 families responded to a survey questionnaire, we know that 30% of girls and women with RTT have bone fractures by the time they reach 20 to 30 years of age. In a follow up cross-sectional study on the natural history osteopenia, we found that bone mineral deficits occur in nearly 50% of females with RTT and these deficits increase with advancing age in individuals with RTT.

Preliminary data from our IRSF funded study titled “Randomized, Placebo-controlled Trial of Oral Calcium Supplementation for Osteopenia in Girls and Women with Rett Syndrome”, suggests that dietary calcium and vitamin D may be associated with the bone mineral status of girls and women with RTT. Both nutrients work together to support bone health; calcium is known to be important for the structural integrity of bones and vitamin D is important in facilitating calcium absorption from the gastrointestinal tract for bone use. From our natural history study, we know that girls and women with RTT consume, on average, only one or two glasses of milk daily -an amount that does not fully support the body’s needs for bone health. We also know that approximately one-fifth of these individuals have vitamin D deficiency when measured by examining blood serum levels of 25-hydroxyvitamin D. Currently, every effort is being made to ensure adequate dietary calcium and vitamin D intake in girls and women with RTT.

What do we want to know?

In order to improve bone health outcomes, we want to know if a dietary calcium supplement will halt the progression of bone mineral deficits and improve bone mineral deposition in girls and women with RTT.

[Link to PDF](#)

Happenings

Featured Angel: Gabriella Leone Peace

Hi! My name is Gabriella Leone Peace but everyone calls me Ella. I am 2 years old and live with my Mommy and Daddy in Carrollton/Dallas, TX. I am at my happiest when there is a big crowd around me. I smile and laugh all the time. I enjoy sitting in my stroller as we walk through malls or around the pond near my house. I also love to watch football and basketball with my Daddy, but my favorite shows are Yo Gabba Gabba and Dora the Explorer. They really make me laugh a lot.

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

Support our first TEAM RETT in the Boston Marathon.

One Mile Closer to a Cure

Thanks to the John Hancock Non-Profit Charity Program, we will have our first TEAM RETT running in the 115th Boston Marathon on April 18, 2011!

All proceeds will benefit the IGF1 clinical trial beginning at Children's Hospital Boston this month! This growth hormone, already FDA approved, will be given to girls with Rett syndrome in hopes that this devastating disorder can be reversed. This clinical trial is giving our families much hope for a better future.

Read about our TEAM RETT members and see why they are running for Rett syndrome.

DONATE NOW

[Learn More](#)

Miss Teen USA Helps Local Girl Fight Rett Syndrome

Abby Diamond (RTT, age 8), daughter of IRSF Board Member, Eric Diamond, was accompanied by special guest Miss Teen USA Kamie Crawford at Chittick School. Kamie was there to visit students and to show her support for Abby and Rett syndrome.

[Check it out on Facebook!](#)

Check out Kamie's blog!

[Read More!](#)

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

Join us at the IRSF Annual Family Conference in Boston, MA. Attendees will meet and learn from top Rett syndrome experts in the field about current care and advocacy strategies, plus hear the latest in research and clinical trials. Of special interest this year will be the opportunity to attend a first-ever full session track on research with leading researchers from Harvard, MIT, and Omar Khwaja, MD, PhD, director of the Rett Syndrome Program in the Department of Neurology at Children's Hospital Boston and principal investigator of the newly begun IGF-1 clinical trial.

Visit the website for newly updated Conference Program and registration information.

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

[Register 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

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](#)

[A Glance @ Rett](#)

[Join a Support Network](#)

[Find your Regional Rep](#)

[Shop](#)

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Past Issues

Tell a Friend

Recent scientific articles related to Rett Syndrome

A Critical Role for Neurofascin in Regulating Action Potential Initiation through Maintenance of the Axon Initial Segment.

Zonta B, Desmazieres A, Rinaldi A, Tait S, Sherman DL, Nolan MF, Brophy PJ.

Neuron. 2011 Mar 10;69(5):945-56.

MeCP2 is required for global heterochromatic and nucleolar changes during activity-dependent neuronal maturation.

Singleton MK, Gonzales ML, Leung KN, Yasui DH, Schroeder DI, Dunaway K, Lasalle JM.

Neurobiol Dis. 2011 Mar 17. (This research was supported by IRSF)

Neuron. 2011 Mar 10;69(5):945-56.

Zonta B, Desmazieres A, Rinaldi A, Tait S, Sherman DL, Nolan MF, Brophy PJ.

Neuron. 2011 Mar 10;69(5):945-56.

Age-dependent expression of MeCP2 in a heterozygous mosaic mouse model.

Smrt RD, Pfeiffer RL, Zhao X. (This research supported by IRSF)

Hum Mol Genet. 2011 Feb 17. [Epub ahead of print]

Brain region-specific expression of Fxyd1, an Mecp2 target gene, is regulated by epigenetic mechanisms.

Banine F, Matagne V, Sherman LS, Ojeda SR. (This research supported by IRSF)

J Neurosci Res. 2011 Mar 10. doi: 10.1002/jnr.22608. [Epub ahead of print]

Social impairments in Rett syndrome: characteristics and relationship with clinical severity.

Kaufmann WE, Tierney E, Rohde CA, Suarez-Pedraza MC, Clarke MA, Salorio CF, Bibat G, Bukelis I, Naram D, Lanham DC, Naidu S.

J Intellect Disabil Res. 2011 Mar 8. doi: 10.1111/j.1365-2788.2011.01404.x. [Epub ahead of print]

Upcoming Events

Natural History Research Study - Chicago

North Carolina Family Picnic

St. Louis Mouse Races

Riverside Rett Lunch And Auction

Georgia Family Picnic

Rett Spring Fling

HER Night - Nevada

Ava's Blues II

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.

Rett Syndrome Natural History Study

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

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

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

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!

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.

In the News

MeCP2: structure and function

De novo MECP2 duplication in two females with random X-inactivation and moderate mental retardation

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 isn't about waiting for the storm to pass. It's about learning to dance in the rain."

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

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