

Rett *gazette*



Winter 2016



Rettsyndrome.org's Advocacy Campaign is a Success!

Rett Syndrome Signed Into Law as an Eligible Condition to Compete for \$278 Million in 2016-2017 Federal DoD Research Dollars

In 2015, Rettsyndrome.org undertook an ambitious advocacy campaign on Capitol Hill with the intent of enabling Rett syndrome research to be an eligible condition in the Department of Defense (DoD) research program. Through the strategic help of Rettsyndrome.org board member Scott Parven, the innumerable families who sent in letters to members of Congress, and the dogged persistence of RSO's Chief Science Officer, Dr. Steve Kaminsky -- the campaign was a success.

On December 18, 2015, President Obama signed into law a bill designating Rett syndrome as one of only 40 areas of study eligible for up to \$278M under the DOD's peer-reviewed medical research program.

This is a time for all of us to rejoice at this new pool of funding available to help advance near-term treatments and ultimately a cure for those with Rett Syndrome. Over the next few months we will be working with Rett syndrome researchers to help them understand this opportunity.

But this is not a time to slow down. The costs of clinical trials far exceed the granting capacity of private foundations. Having Rett syndrome included in this bill will allow researchers to apply for this pool of funding from the DOD which will help with clinical trial funding. **As a community, this is the very time to expand our efforts to accelerate our research programs.**

Why is Rett Syndrome included in DoD funding?

Both Rett syndrome and Traumatic Brain Injury (TBI), which affects a number of wounded warriors, have associated synaptic dysfunction. In searching for treatments for Rett syndrome, researchers may uncover a host of pathways to treat the TBI epidemic and other neurological dysfunction that inflict veterans—including Parkinson's Disease and Alzheimer's.

What does this mean for the research community?

Researchers investigating compounds to treat Rett syndrome can apply for DoD funding to support their research under this program.

What does this mean for families?

Lawmakers have recognized the importance Rett syndrome research plays in gaining a better understanding of many neurologic disorders. Most importantly, this appropriation provides a significant new funding opportunity for federal research dollars towards the study of treatments for Rett syndrome. We hope you are as encouraged by this legislation as we are. We are and will continuously look for ways to expand research and increase the value of your critical donations to Rettsyndrome.org in our goal to help all children affected by Rett syndrome.

Participating in a Clinic Trial?

With two promising clinical trials underway, we know that many families have questions about participating. This topic is especially timely, because our Board of Directors has approved \$439,000 more for the Phase II IGF-1 trial and \$750,000 more for Neuren Pharmaceutical's Phase II trial of trofinetide. This combined funding of nearly \$1.2 million will allow:

- The Phase II IGF-1 trial to recruit 17 more people and expand to two additional sites, the Greenwood Genetic Center in Greenwood, South Carolina and Rush University in Chicago.
- The Phase II trial of trofinetide will study a pediatric population of children between age 5 to 15, and it will also be a multi-site trial. Neuren Pharmaceutical's has announced the initial list of participating sites. Visit www.rettstudy.com to see the listing and stay updated on trial announcements. Equally important, the new pediatric data will supplement last year's findings regarding trofinetide's impact on females age 16 and older.

An Interview with the Boom Family

To help you learn about clinical trials, we interviewed the Boom family of Deerfield, New Hampshire. Parents Greg and Michelle Boom enrolled their 5-year-old daughter, Isabelle (Izzie) in the IGF-1 trial.

How did you learn about the IGF-1 trial?

When we got the results of Isabelle's genetic testing from Children's Hospital at Dartmouth, we also learned that Boston Children's Hospital (BCH) had a Rett clinic. It was during our first visit to the clinic that BCH's Dr. Kaufmann told us about the trial and that Isabelle would be a great candidate.

What is it like being involved in a clinical trial?

From April 2014 to August 2015, we had to make six visits to BCH. Each visit required 7 days of different screenings, learning tests, an EEG, and an EKG. Our appointments always started at 8 a.m. Since we live close to BCH, we never stayed in Boston like most families. Most days we were there for the whole day, especially if Izzie wasn't having a good day. But there were some days when our visits only required 2 hours. In addition to the visits, there were 8 telephone check-ins with BCH that involved questionnaires.



The Boom family includes Greg and Michelle with their children Isabelle (5), Carter (3) and Tyler (11 months).

Parent, Michelle Boom's thoughts for families considering a clinical trial

You have nothing to lose! If you ever have an opportunity to give your daughter a fighting chance at her life, why not jump all in and see what can happen? You will never want to look back and think what if – what if you let the opportunity slip by? At least if you gave a trial a shot, you will have no regrets.

Our Sincere Thanks

Thank you to all past, present and future families who participate in clinical trials for Rett syndrome. You are paving the way for advancements in research. We know the road isn't easy, but we are grateful for all the dedication and hard work you have done to support the community and the mission of our foundation.



New Gala Event Raises Nearly \$1 Million

On November 18th we held a new gala event – Reach for the Stars. The Boston event honored Jane and Michael Joyce, two extraordinary champions of Rett syndrome. Over the years, the Joyces have hosted countless fundraising events for Rettsyndrome.org in honor of their daughter with Rett syndrome, Caroline. These events not only resulted in significant funding for our mission but they also raised awareness of Rett syndrome throughout the New England area. We are honored and grateful to have the Joyces among our Rett community.

Reach for the Stars will become one of our annual gala events, and much of this year's success is the result of the hard work and dedication of our newest board member – Kim Jacobs. Kim and her husband Charlie chaired the event, and the Jacobs founded the Boston Bruins Foundation. The Jacobs are very committed to community service, and they dedicate much of their philanthropic investments for at-risk children and/or children with medical needs. Like our champions, the Joyces, we are also very grateful to the Jacobs for joining our collective efforts.

Rebel Against Rett

The Rebel Against Rett event was hosted June 27th in Chicago. The two part event involved a strength challenge in the morning and a party in the afternoon and was planned by Mike Connelly and Amy Zima. \$17,000 was raised during the event. We thank Mike, Amy and everyone that participated in the event for raising awareness and funds for Rett syndrome!

Volunteers are at the core of Rettsyndrome.org. Without our volunteers, who host hundreds of events each year, we would not be able to continue funding research to better understand, treat, and ultimately find a cure for Rett syndrome. Thank you to all that have hosted or attended events in 2015!

Strollathons

The 2015 Strollathon season has officially come to a close. There were a total of 26 Strollathons that took place in 2015 and out of those two were new – Southern Texas and Delaware.

We would like to recognize the top 10 individual fundraisers for the 2015 Strollathon season. It is truly impressive what one person can raise!

Angel Espinosa • Elizabeth Allocco • Anne Rich
Sandy Kopelman • Lily Perez • Jacy Horst • Tracy Bergman
Paula Bell • Karli Hughes • Jennifer Endres

**Thank you to
everyone that raised
funds for Rett
syndrome in 2015!**

Strollathon
Stroll & Roll for Rett Syndrome



Rett Racers

Thank you, Kevin Streifel for running the Ragnar Relay in Minnesota as a Rett Racer. Kevin and his wife, Amber raised \$5,000 to support Rettsyndrome.org in honor of their daughter, Cora! As Kevin explained, "We run with her in our mind and soul. We run because she hasn't learned yet, and we know that smile and giggle is her love and motivation for us to be her feet until she can." Consider joining Kevin as a Rett Racer by signing up at www.retracers.org.

Investigator Spotlight – Meet One of Our Researchers

Behind every clinical trial and advancement in research are dedicated and hard-working scientists, doctors, and researchers. Rettsyndrome.org appreciates these investigators' work and their personal commitment to helping us learn more about Rett syndrome. Of course, these researchers also identify treatments and assistive devices that help improve the quality of life for those living with Rett syndrome.



We want to bring these men and women out of the labs and into the spotlight so that our community gets to know them better. Today our spotlight is shining on Dr. Jeffrey Neul of the University of California at San Diego (UCSD). This year, Dr. Neul was awarded one of our ANGEL grants – a \$600,000 grant that is allowing him to identify new compounds and drugs that might be promising for Rett syndrome. He then tests his findings in animal models.

What led you to your career as a researcher of Rett syndrome?

I became interested in Rett syndrome during my final year of medical school. When I asked my child neurology mentor, Peter Huttenlocher, where I should train for child neurology, he picked up an open journal lying on his desk and said: “Huda Zoghbi identified the gene for Rett syndrome, so you should go work with her.” It was one of the best pieces of advice any one has ever given me.

What is the most rewarding aspect of conducting Rett syndrome research?

I love working with the children affected with Rett syndrome and thinking about how we can do research to help them live fulfilling lives.

If you could pick any one symptom of Rett syndrome to prevent or to provide relief for, what would it be?

If I could, I would try to figure out how to let these children talk. I know they have a lot of things they want to tell their families, and being able to communicate would make a remarkable change in their lives.



Investing in Rett Syndrome Research

Our thanks goes to Robert Costantino, who made a passionate plea for Rett syndrome to a group of investors at a Las Vegas celebration. To date, his effort has resulted in over \$55,000 for Rettsyndrome.org, in honor of his neighbor's daughter, Hannah Varbel. We greatly appreciate the generosity of Rob and all those who stepped up in a big way to help!

Climbing Mt Kilimanjaro for Rett Syndrome

On August 11th, Maria McEvoy reached the summit of Mt. Kilimanjaro – the highest mountain in Africa - in order to raise funds and awareness of Rett syndrome.

It was a goal she set for herself to help commemorate her 50th birthday and to pay tribute to her niece, Emma. She had hoped to raise \$19,000 – one dollar per each foot of Kilimanjaro, but Maria far surpassed that goal having raised \$33,500 to date! Thank you, Maria!

Meet One of Our Dads – Shannon Santos

There is a special bond between dads and their daughters. Our Rett community dads inspire us with the love and care they show their daughters. Each month we honor one of these special dads by featuring them on our website and social media. This month, we are honoring Shannon Santos for all his love and support of his daughter, Melaina. You can also read about other dads like Shannon by visiting our website Rettsyndrome.org. If you would like to be featured as Dad of the Month, please email Family Resource Manager, Jennifer Endres at jendres@rettsyndrome.org. We'd love to hear from you!

Shannon's Unconditional Love for Melaina

I can remember the days before Rett syndrome. I remember holding Melaina for the first time as a newborn and having her lay next to me as she loved to snuggle. I remember coming home from work and hearing "Daddy home." I remember the stories from my wife when Melaina would playfully call back as they drove from grandma's house "Going to the store" and "Horsey."



I also remember Melaina playing tag with her brother in the hallway and diving on the floor. I remember her sliding down the stairs on her belly and climbing them and doing the same thing over and over. Then I remember the delays in speech, the loss of "Daddy home," the hand mouthing, the hand wringing behind the back, the arm flapping, the pincer grip, teeth grinding, toe walking, walking aimlessly, and the worry of something wrong. This something wrong turned into "Wait and see" from doctors. She's still developing." This instinct of something is wrong also turned in to 48-hour EEGs, EKGs, CAT Scans, and blood tests. I remember the "Owie!" during the blood tests. I remember the seizures, the ambulance rides, the hospital stays, and all these things that I now want to forget.

Then I think about what my wife and I have. Melaina is a beautiful, smart, and intuitive seven-year old, second grader who loves Bruno Mars, The Backyardigans, fried foods, snack foods, swimming, and biking. A wheelchair, an adaptive bike, therapy (speech, physical, hippotherapy), and a Tobii are among those things that have given us solutions and ideas on how to deal with all the challenges that Rett syndrome presents Melaina. We can only hope for what the future holds for many girls affected by Rett syndrome.

I lost who I was as a husband, father, brother, son, and friend only to find that I needed to accept the challenge of finding a silver lining and find joy as my family and friends deserve a healthy, better me. Melaina is here to teach more than "that a life with Rett syndrome sucks." She is here to teach me about compassion, unconditional love, patience, hope – all of which helps us face challenges in the darkest of times.

Golf Outings

2015 marked a big year for Golf Outing, which raised over \$618,000 for Rettsyndrome.org. The Samantha Corpus Golf Outing celebrated its 10th anniversary in May. This annual event held in Blackhawk, California has raised over \$1.7M dollars since it started. We thank the Corpus family for their dedication to finding a cure for Rett syndrome.

Stronger Together

As a rare disease organization, we know the benefit of partnering with other organizations dedicated to MECP2 research. Last year's new NIH National Center for Advancing Translational Science (NCATS) Rare Diseases Clinical Research Network (RDCRN) funding for the Natural History Study demonstrates their belief in studying related disorders as well.

Rettsyndrome.org is now an active member of that network – specifically the Research Consortium of Rett syndrome, MECP2 Duplications and Rett-related disorders. The Consortium is led by Alan K. Percy, M.D. (University of Alabama at Birmingham) and involves clinical research into four related disorders of the nervous system: Rett syndrome; MECP2 Duplication disorder; CDKL5 disorder; and FOXP1 syndrome.

In 2016, we will invest \$65,000 to expand the Natural History Study to three more locations. Our goal for this investment is to ensure wider geographical access so more families will consider participating in the Study. Ultimately, this research project allows us to learn more about Rett syndrome and its course of progression. Early next year, as a result of our \$65,000, these three new sites will begin enrollment in the study:

1. Gillette Specialty Healthcare in Minneapolis
2. The Children's Hospital of Cincinnati
3. Washington University in St. Louis

If you're interested in enrolling in the Natural History Study, please contact our Director of Training and Development, Paige Nues at 1.800.818.7388 or at pnues@rettsyndrome.org

Unity Strength and Hope – 2016 Family Conference & Research Symposium

We are partnering with our Natural History Study Consortium members for the 2016 Family Conference. The conference's theme is Unity, Strength and Hope and will be June 22nd – 24th at the Eaglewood Resort in Itasca, Illinois. This will be a wonderful way to promote Rett syndrome research, empower families, and broaden our rare disease perspective. It is through collaboration and discussion that we move closer to treatments and best-care practices for those with Rett syndrome. Our Consortium partners (The International Foundation for CDKL5 Research; MECP2 Duplication; and the International FOXP1 Foundation) are equally committed to a successful conference for families.

Immediately preceding the conference, we will host the 15th Rett Syndrome Research Symposium. The event brings together researchers, clinicians, therapists, allied health professionals, and research advocates for three days of talks on topics such as clinical research, basic and translational research, neuro-habilitation research, and the Natural History Study. Attendees who register for the Family Conference are invited to attend the Friday afternoon Research session on the Natural History Study.

To keep registration fees reasonable for families, we are seeking sponsors for the conference. If you or your employer would like to be one of the conference's sponsors, please contact our Director of Training and Development, Paige Nues at 1.800.818.7388 or at pnues@rettsyndrome.org.

UNITY * STRENGTH * HOPE
RETTSYNDROME.ORG • EAGLEWOOD RESORT, ILLINOIS • 2016
RESEARCH SYMPOSIUM (JUNE 22-24) • FAMILY CONFERENCE (JUNE 24-26)

UNITY * STRENGTH * HOPE
JUNE 24-26 • EAGLEWOOD RESORT, ILLINOIS • 2016
RETT SYNDROME, MECP2 DUPLICATION, CDKL5 DISORDER, FOXP1 DISORDER
FAMILY CONFERENCE

Attend two events for the price of one! It's a great year to attend the family conference, because your registration also allows you to attend the Abilities Expo –which is being held during the same days as our conference. Check it out: abilities.com/Chicago for details about the hundreds of product companies and service providers that will be on site.

Meet New Board Member, Kim Jacobs

Our success begins with the right people working together for the same cause – Rett syndrome. In order to accomplish our important work, our Board of Directors continues to grow and secure the right mix of people throughout the country who will lead us into the future.

We're pleased to welcome our newest board member, Kim Jacobs. Kim brings an extensive history of philanthropic history and involvement with causes that benefit children with medical needs. Kim and her husband, Charlie founded The Boston Bruins Foundation - to assist other charitable organizations that demonstrate a strong commitment to enhancing the quality of life for children in the community. The Foundation has provided millions of dollars to countless community organizations, including a gift of \$50,000 to Rettsyndrome.org. We are thankful to Kim and The Boston Bruins Foundation for their commitment to Rettsyndrome.org. We also look forward to working with Kim in the years ahead.



Stay Connected & Informed – Our Website Is for You!

We've launched an incredible web-based resource for our Rett community. This newest project, What's in Your State, helps our Rett community quickly learn about specific resources in their state ranging from accessing Waivers to In-Home Support Services or Family-fun Leisure Destinations. To date, family volunteers have helped create content for eight U.S. states, and we will be finalizing the rest throughout 2016. All of the resources are organized by state and are available in one easy to find place. There is even a listing of resources that are common across all 50 states. Be sure to check them out by visiting www.rettsyndrome.org/for-families/resources.

One common resource that many states have are Rett Syndrome Associations. These Associations serve as important support systems for families of individuals living with Rett syndrome. The Associations are also some of our most important partners, helping us to raise funding and awareness of Rett Syndrome.

One such example is The Midwest Rett Syndrome Foundation. Established in 2002, the Foundation has grown from its original support of Minnesota families and now serves families living throughout the Midwest. If you are a Midwest family and would like to learn more about the foundation, please contact Lisa Evert at lisa.evert@mrsra.org to find out how to get involved.

For New England families, please don't forget about your resource - the Rett Syndrome Association of Massachusetts (RSAM). Each year RSAM's Boston Marathon Team of Rett Racers support us by raising money and building awareness of Rett syndrome. This year alone, RSAM donated \$85,000 to support the ongoing clinical trial of IGF-1 at Boston Children's Hospital. If you are interested in joining the Rett Racers who participate in the annual Boston Marathon, or if you want to get involved in the Rett Syndrome Association of Massachusetts, please contact Jane Joyce at jane.joyce@comcast.net.

October Awareness Month

October was again our month to raise awareness of Rett syndrome and its impact on children and adults. This year we teamed up with 16 other Rett organizations from around the world to share WHY people give to Rett syndrome. Throughout October, stories poured in from Western and Eastern Europe, Asia, the Middle East, Latin America, and North America. It was an amazing way for all of us to express our collective impact and tremendous efforts on behalf of those living with Rett syndrome. Thank you to everyone that participated in the Why Would You Give campaign!



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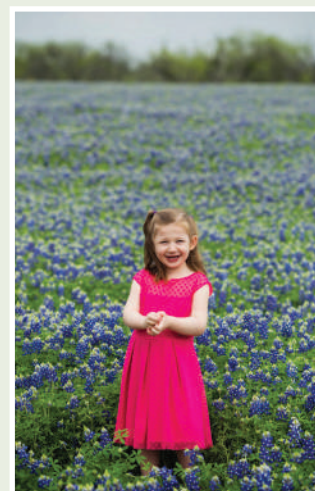
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Every effort has been made to ensure that this list is accurate. We apologize if we have inadvertently missed a name.