

# Rett *gazette*



Summer 2015

## Your Help Is Our Hope

*A special thank you to our generous volunteers*

It has often been said that volunteers are love in motion. We are fortunate to have a huge community of volunteers who support our mission and put their love of girls with Rett syndrome into action. Below are just two events of many that highlight our volunteers' creative and tireless fundraising and advocacy efforts on behalf of our cause, our community and, most importantly, our girls!

### Walk the Lake for Blake

One volunteer whose efforts represent "love in motion" is grandfather, Kevin Goodman. When his granddaughter, Blakely, was recently diagnosed with Rett syndrome, he and coworker, Jeanetta Wilson, organized a creative fundraising event — Walk the Lake for Blake. It was held at The Boeing Company's Huntsville, Alabama campus.

Boeing donated \$100 for every Boeing employee who participated — ultimately raising over \$20,000. We greatly appreciate Boeing's generosity and the efforts of all who participated. Special thanks also to the Goodman family, who hope to continue this creative event annually.

### Color Run

Finding new and inventive fundraising events is always one of our goals. Last year, Nicole Karnash, parent of 19-year-old Noelle, suggested a great new idea — The Color Run. These new 5K runs are sweeping the nation, and Nicole helped us jump on board. In fact, we were the official sponsor of last year's Hershey, Pennsylvania Color Run — which raised over \$80,000. The event was so popular that this year it will be



*The Hershey, Pennsylvania Color Run raised over \$80,000.*

held two days. It will be exciting to see how it grows in the future, and we look forward to partnering with Color Runs in other cities. We appreciate all of Nicole's help.

## Cammy Can

Many of you "met" the Babiarcz family and their amazing daughter, Cammy, in our recent appeal letter. They not only manage to find some positive aspects of life with Rett syndrome but they also share the generosity they've received from others with our larger community. One example is their annual family fundraiser, Cammy Can's Cinderella Story. This year's event raised necessary money for our important work — including \$1,300 from the sale of Cammy's original art work. When your



time permits, we hope you will read about Cammy in our spring appeal and consider making a contribution that will move us

closer to a cure. Please visit <https://www.rettsyndrome.org/get-involved/spring-appeal> to donate today.

# We're Investing \$2.9M for Research

Last year we funded 22 new grants and launched the Neuro-Habilitation Program to develop cognitive therapies, physical therapies, occupational therapies and speech therapies for skill development in Rett syndrome. As always, we selected our research investments based on a rigorous peer-review process. These investments align with our strategy of aggressively moving more potential drugs to clinical trials while also identifying treatments.

One example is the ongoing positive developments about Neuren Pharmaceuticals' Trofinetide: Last November, Neuren announced its Phase II clinical trial results — sharing the optimistic news that Trofinetide was safe, well tolerated, and effective among the higher dosage group of girls and women, ages 16 to 45, receiving Trofinetide.

In February, there was more good news from Neuren: the FDA granted Orphan Drug Status to Trofinetide. The Orphan Drug designation means (among other things) that Neuren has the exclusive intellectual property rights for researching and manufacturing Trofinetide. This is good news for families and a historical breakthrough! For the first time ever, identifying a treatment for Rett syndrome is a priority for a pharmaceutical company and the FDA.

## What are Neuren's next steps?

In the coming months, we anticipate Neuren will propose another Phase II trial. This trial might target children ages 5 to 10 years old, but no final decision on age range has been made at this time. It will be of similar size as the last trial and is likely to occur at more than one site. If the trial is found to be effective, it could lead to a Phase III trial (Pivotal Trial) to determine if Trofinetide is, indeed, an effective treatment for Rett syndrome.

## IGF-1 Update

Later this year, the IGF-1 trial will expand to a second site — the Greenwood Genetic Center in Greenwood, South Carolina. The trial's expansion beyond Boston Children's Hospital was the recommendation of the FDA when the study's lead researcher, Dr. Walter Kaufmann and Ipsen Biopharmaceuticals recently met with the FDA. Our Chief Science Officer, Dr. Steven Kaminsky, also attended the meeting as both a patient advocate and because Rettsyndrome.org is the trial's current sponsor. The FDA meeting's purpose was to discuss treating Rett syndrome patients with Increlex® (IGF-1) for social-avoidance behaviors and anxiety-like symptoms. Increlex® (generic name, Mecasermin) is a

human insulin-like growth factor that has been marketed in the US since 2005 for the indication of severe short stature due to IGF-1 deficiency or growth hormone gene deletion. Since Dr. Kaufmann's Phase II clinical trial demonstrated IGF-1's effectiveness for addressing some quality of life indicators for those diagnosed with Rett syndrome (such as social avoidance and anxiety), the meeting with the FDA sought guidance about the appropriateness and acceptability of studying social avoidance as a primary outcome for Phase III clinical trial.

In the short term, however, the FDA recommends that a Phase II trial be conducted at two locations in order to avoid possible researcher bias. Such a recommendation is typical when seeking FDA approval for a new indication of an existing drug. Consequently, we're investing additional funding for the trial's expansion to Greenwood Genetic Center. While the FDA's recommendation means a delay for moving to a Phase III trial, it will ultimately result in a more comprehensive clinical trial. Details about the expanded study are being worked out, and more information will be shared later this year.

## Research to Reality Campaign

2014 was an extraordinary year for the Rett syndrome community. Within just 12 months, we went from anxious uncertainty to affirmed reality — the reality that the Phase II clinical trials of both Trofinetide and IGF-1 are finding possible treatments for some symptoms.

But these two potential treatments aren't a cure. So last fall, we launched the Research to Reality (R2R) Campaign. To date, our board members have contributed over \$1.5 million towards this year's goal of \$5 million. And our hope is to raise \$100 million during R2R's multi-year campaign.

Every dollar raised will only be used for research! For instance, this year's \$5 million will help to advance the Trofinetide and IGF-1 clinical trials — while also funding research of more than 20 promising compounds.

**You too can help translate research to reality.** Please consider visiting our website to invest in the R2R Campaign. Together we can move research from the lab to the reality of treatments and a future cure.



# Out & About for Rettysyndrome.org

Depending on what part of the country you live in, many of you have already been “out and about” raising money for our important work. Others of you are just gearing up for your special events. We thank you all! Last year, your generous efforts helped us to raise a total of \$2,344,520 from special events. Below is just a sample of some of 2015’s upcoming events and a special callout of gratitude to those of you who have already held events this year.

## Louisiana Marathon Raises Over \$86,000

In January, runners from across the state of Louisiana, including many from the Albemarle Foundation, crossed the finish line of the annual Louisiana Marathon. Our Rowdy Rett Racers ran with smiles and waves while supporters stood at the 9-mile marker cheering them on. Special gratitude goes to Kathryn Kissam and the Albemarle Foundation for their hard work and successful fundraiser for many years now!



*Over \$86,000 was raised by runners and supporters in the Louisiana Marathon.*

## Golf Benefit in Honor of Charlotte Calderazzi

Larry and Pat Calderazzi come from a small Florida community with some great people. Their granddaughter, Charlotte, has Rett syndrome, and many of their friends and neighbors attended their Tee’s & Rett’s Closest of the Pin Golf Benefit. The event not only raised awareness of Rett syndrome, but it also raised \$1,250. Charlotte’s parents then matched the amount — bringing the total to \$2,500. We greatly appreciate the generosity of the Calderazzi family and those who supported their efforts.

## Love for Leila

House 4 Humanity served up a fun and memorable evening for Leila, age 4, and her family. House 4 Humanity is a San Francisco charity that draws from the music scene to fundraise and raise awareness of children suffering from illness, hunger, and homelessness. They hosted a “Love for Leila” event at a new San Francisco music spot that raised over \$4,500 for Rettysyndrome.org and helped to build awareness of Rett syndrome. Thank you!



## 2015 Spring Strollathons

Get ready! Strollathon season has begun, and it’s time to Stroll & Roll for Rett. Below is a listing of some upcoming Strollathons. You can also check out [www.strollathon.org](http://www.strollathon.org) to find a strollathon near you. Each year, the people who Stroll & Roll for Rett raise significant money for our important work. We thank all of you!

**St. Louis Strollathon** – Saturday, May 2nd – Joyce Opinsky and Amy Kreher

**Chicagoland Strollathon** – Sunday, May 17th – Shelby Swann

**Michigan Strollathon** – Sunday, May 17th – Lisa Fenberg

**Delaware Strollathon** – Saturday, June 13th – Kenna Seiler

# Meet One of Our Angels: *Ava Guy*

We like to regularly spotlight one of our many inspiring Rett angels. Below we feature angel, **Ava Guy**. You can read about other angels like Ava by visiting our website [Rettsyndrome.org](http://Rettsyndrome.org).

Hi! My name is Ava and I am four and half years old. I am very lucky to have a loving family that takes care of me, including my mom, dad and two sisters. I also have an amazing bond with my nana. All people I come in contact with love my infectious smile and courageous attitude. They know that I dislike lateness and love having a routine. Other things I like include my dog, bubbles, fireworks, and macaroni and cheese. My family learns from me every day and they say that I am an inspiration to them!



## Angel Awareness Cards

Tell the world about your angel with Rett syndrome with our [Rettsyndrome.org](http://Rettsyndrome.org) personalized Angel Awareness Cards. You can request your cards at [Rettsyndrome.org](http://Rettsyndrome.org) — then print and share them with your community.





## New Publication – The First 100 Days Pathway

We have created a new resource, ***The First 100 Days Pathway***. Its purpose is to help families who have recently learned that their child has Rett syndrome. One of its benefits is helping families organize their “To Dos,” so to speak, by prioritizing suggested action steps for the first 30, 60 and 90 days. Covered topics and related action steps include: family and community; medical considerations and assistance; potential therapies; and current research.

Created by families and our professional advisors, *The First 100 Days Pathway*

reflects our commitment to provide timely and relevant support to families — whether on the day of diagnoses or throughout their experiences with Rett syndrome. Essentially, it’s a “one-stop” resource at a time when parents are most desperate for a comprehensive resource that allows them to know where to begin with meeting their child’s needs.

Please contact us for a free copy. We’re here to help you.

## Mark Your Calendar for June’s Family Conference



Rather than host a traditional conference that only a few hundred parents can attend, this year’s family conference will be virtual. Since traveling is difficult for so many families, the goal of our virtual conference is to ensure your access to presentations on recent research, new therapies, various symptoms, and other important topics for daily life with Rett syndrome. Additionally, the conference’s numerous presentations will be available for later viewing, via our website.

If you are able to attend the virtual conference “live,” please mark your

calendars for June 22nd through June 26th. The daily schedule and online registration will open June 1, 2015. Visit <https://www.rettsyndrome.org/for-families/family-conference> for more information!

Next year, we will return to our traditional conferences. Both the Rettsyndrome.org Science Symposium and Family Conference will be in the Chicago area on:

**June 22 - 24, 2016** – The Science Symposium

**June 24-26, 2016** – The Family Conference

## October’s Family Getaway - Orlando!

While our June conference will be virtual, we know the value of bringing families together for support, friendship, and shared learning.

So please join us the weekend of October 2nd in Orlando! There are no meetings or registration fees — just a chance to be together and spend time with our extended Rett syndrome family!

Details and special room rates at the Disney Port Orleans can be found at [www.mydisneymeetings.com/rett-syndrome-family-getaway](http://www.mydisneymeetings.com/rett-syndrome-family-getaway).

***Don’t miss October’s family getaway to Orlando, Florida!***



# Memorials

The loss of every child is a loss for our entire community. Please join us in a moment of reflection for all of the beautiful children who have gone before us, and especially for these families who have asked for our support during their recent loss. We promise to learn from each and every one of them, remember them always, and allow their lessons to guide our future efforts. We share in these families' tremendous heartbreak:

Kristen Jean Allee, FL, age 24

Giselle Anaya, CA, age 16

Stephanie Baer, CO, age 24

Kristen Bean, WV, age 24

Megan Beard, TX, age 22

Kianna Bisquera, CA, age 14

Kelsey Boedigheimer, AK, age 21

Ella Caroline Boll, Alberta, age 15 mos.

Nancy Brown, MI, age 56

Gia Nicole Cail, FL, age 3

Jordan Caron, ME, age 10

Kate Chrzan, NY, age 20

Marianne Cino, NJ, age 40

Allison Cowan, NY age 19

Kaitlyn "Amber" Gant, AL, age 19

Liora Goldberg, Israel, age 14

Amber Heiss, OH, age 13

Skylar Hopper, NJ, age 8

Alyssa Horan, IL, age 12

Gabrielle Jankowski, NJ, age 11

Chelsea Jauch, MI, age 16

Nie' Jones, SC, age 12

Lily Kaelin, WA, age 4

Sara Kaltmayer, IL, age 15

Taylor Kramer, PA, age 22

Charlotte (Charlie) Lossner, NV, age 28

Jessica Marsillet, KY, age 29

Olivia Fay Miller, MN, age 11

Samara Bella Peniston, TX, age 5

Ashley Ritter, IL, age 38

Jaclyn Ropiecki, NY, age 27

Marissa Sanchez, CA, age 28

Abigail Shack, IL, age 12

Allyson Stromberg, NJ, age 20

Renee (Nee' Nee') Thompson, MN, age 30

Sarah Turner, RI, age 24

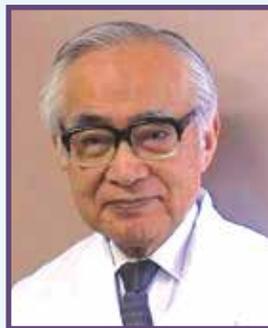
*These Memorials from 2009 – May 2015 were recently received. Please visit our website to read full Memorials in support of these families.*

## Life After Rett

We encourage families who have lost someone to Rett syndrome to join our "Life After Rett" Facebook group. This is a monitored group, so you must request to join it. But it is a place to find support and sympathy from those who understand your loss and grieving process. Members support each other and offer encouraging ways to commemorate loved ones. Rettsyndrome.org sponsors "Life After Rett," and it is moderated by parents Jane George (jgeorge@globetrotter.net) and Elaine Dolecheck (cowgirlandidee@comcast.net).

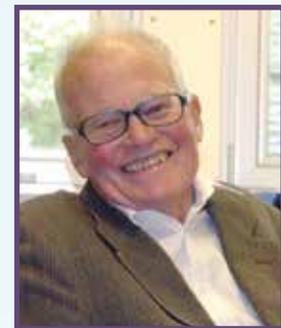


## We Have Lost Two Pioneers



**Masaya Segawa MD, PhD**

It is with heavy hearts that we share news about the loss of two of Rett syndrome's earliest researchers. Last December, we lost **Masaya Segawa MD, PhD** (1936-2014) — one of Pediatric Neurology's earliest researchers. And in April, **Bengt A. Hagberg, MD, PhD** (1923-2015) passed away. In the 1960s, both physicians were credited with establishing the field of Child Neurology as a distinct discipline from general Pediatrics and adult Neurology.



**Bengt A. Hagberg, MD, PhD**

Dr. Hagberg is also credited as one of the first physicians to recognize Rett syndrome in his young patients. In 1960 he began noticing that some of his young female patients were making repetitive hand-washing motions. By 1983, Dr. Hagberg and his colleagues published a breakthrough article in *Annals of Neurology* that resulted in Rett syndrome's recognition by the medical community. Dr. Hagberg chose to name the disorder "Rett syndrome" in honor of Dr. Andreas Rett's early research.

# From Knowledge Will Come a Cure: Organ and Tissue Donations

Organ and tissue donations are one way that families can help researchers unravel the mysteries of Rett syndrome. You can help us learn more by donating a sample of your child's blood or tissue the next time she has a routine blood test or undergoes a medical or surgical procedure — such as a

skin or muscle biopsy, endoscopy, scoliosis surgery, etc.

Naturally, the most difficult thing to consider during times of good health is organ donation. But organ donations are an important gift of hope for discovering treatments and a cure for Rett syndrome. We respectfully

request that U.S. and Canadian families consider this selfless and courageous gift. Please know that the donation process must begin within hours of passing and before any preparations.

If you would like to learn more about organ and tissue donations or our important work with the Harvard Brain Tissue Resource Center (HBTRC), please contact Paige Nues ([pnues@rettsyndrome.org](mailto:pnues@rettsyndrome.org)).



**You can help us learn more by donating a sample of your child's blood or tissue the next time she has a routine blood test or undergoes a medical or surgical procedure.**

**According to Dr. Story Landis, retired director of the National Institute of Neurological Disorders and Stroke:**

*"While making a gift of a loved one's brain to science may seem unsettling to many of us, donors are making an important contribution in advancing knowledge about the structure and function of our most complex organ."*

## Stay Connected & Informed – Our Website Is for You!

Nearly every week, we issue important information on our website. If you haven't visited recently, listed below are just a couple new additions. Also, don't forget you can join a Special Interest Network on our website. These networks provide support during every stage of the Rett syndrome journey. They include groups for dads, grandparents, educators, and several others. You can join one of our Networks at [rettsyndrome.org/for-families/get-support](http://rettsyndrome.org/for-families/get-support).

### Rett Syndrome and Sleep

This is a wonderful resource filled with Frequently Asked Questions pertaining to Rett syndrome and sleep. It was prepared by Dr. Daniel Glaze — Medical Director

of The Blue Bird Circle Rett Center, Texas Children's Hospital. You can find it on our "Typical Symptoms" web page.

### New Rett Clinic

In January, The Rett Spectrum Clinic at Washington University and St. Louis Children's Hospital opened its doors. Read all about this new clinic by visiting our "Clinics" web page.

**Visit [Rettsyndrome.org](http://Rettsyndrome.org) for important information, resources and support during every stage of the Rett syndrome journey.**





## THANK YOU TO OUR PARTNERS FOR MAKING OUR WORK POSSIBLE

### We thank the following groups who have supported us January – May 2015:

#### ER Heroes (\$50,000+)

International Foundation for CDKL5  
Research

#### HER Knights (\$25,000 - \$49,000)

The Leroy Schecter Foundation, Inc.

#### HER Angels (\$10,000 - \$24,999)

Barack Ferrazzano Kirschbaum & LLP  
The Boeing Company  
Brookfield Financial Properties, LP  
Casting 4 A Cure  
The Huntingdon Valley-Churchville  
Rotary Club  
IMC Chicago Charitable Foundation  
J. Calnan and Associates, Inc.  
Manhattan Beer Distributors, LLC  
Newmark & Company Real Estate, Inc.  
Rochester Area Community Foundation  
Rocky Mountain Rett Association  
Seyfarth Shaw Attorneys, LLP  
Southern Ionics, Inc.

#### In-kind Contributions

Akin, Gump, Strauss, Hauer & Feld, LLP  
Attention Span Media  
Baylor University  
Boston Children's Hospital  
Reading Rock  
Seyfarth Shaw LLP  
University of Alabama at  
Birmingham  
University of California  
San Diego  
Wiley Ryan

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

