

See P. 5

# Rettgazette



Spring • 2016

## We Are Leading the Revolution in Rett Syndrome Research

### Neuren Trofinetide Trial Moving Forward

The Rett-002 study of trofinetide is now open for enrollment! Five of the 11 sites are now enrolling participants for the study which will look at a pediatric population of children between age five to 15. This Phase 2 study is truly groundbreaking as it is the largest multi-center trial in Rett syndrome ever conducted. We thank the team at Neuren for their dedication to finding treatments for Rett syndrome.

The sites currently enrolling participants include:

- University of Alabama, Birmingham
- Boston Children's Hospital, MA
- Baylor College of Medicine, TX
- Greenwood Genetic Center, SC
- Rush University Medical Center, IL

At the time of this printing, these were the sites that were open. Visit [www.rettstudy.com](http://www.rettstudy.com) for the most recent details.

### NHS Sites Now Enrolling

Rettsyndrome.org is proud to continue to be a co-sponsor and administrative coordinator of the NIH Natural History Study that was formed back in 2014. The objective of the Natural

History Study remains the same: to gather as much data as possible about children and adults with Rett syndrome. This historical and physical examination data will stimulate more clinical research and better prepare us for future clinical trials. Twelve clinical sites of the 14 have been announced as enrolling and conducting study visits.

Enrolling sites include:

- University of Alabama, Birmingham
- Children's Hospital Boston, MA
- Children's Hospital of Philadelphia, PA
- Vanderbilt University, Nashville, TN
- Baylor College of Medicine, Houston, TX
- Gillette Children's Specialty Healthcare, St. Paul, MN
- UCSF Benioff Children's Hospital of Oakland, CA
- Greenwood Genetic Center, Greenwood, SC
- University of Colorado, Denver, CO

For enrollment contact information and study details, visit [rettsyndrome.org/research/natural-history-study](http://rettsyndrome.org/research/natural-history-study)



## Rett Syndrome Researchers Now Applying for DoD Funding

As you may remember, late last year Rett syndrome was designated as one of only 40 areas of study eligible for up to \$278 million of funds under the Department of Defense Peer Reviewed Medical Research Program. This new pool of funding will help advance near-term treatments and ultimately

a cure for those with Rett syndrome. Researchers investigating areas of research from discovery science to clinical trials and technology and therapeutic developments to treat Rett syndrome can now apply for this DoD funding. The government has now opened up five programs to

pre-applications. In the coming months researchers will begin the application process. We look forward to supporting these researchers and their critical work.

For more information visit [www.retttsyndrome.org/blog](http://www.retttsyndrome.org/blog)

## Working Together to Raise Money

### *A special thank you to our generous volunteers*

*Each year people all across the country host fundraisers and send out special appeals to their families and friends in support of Rettsyndrome.org. We appreciate the support of all fundraisers, whether big or small, as they all go to help Rettsyndrome.org achieve our mission. Below are just two of the many examples of the tireless efforts of our volunteers.*



*Colleen Cler*

### 10th Annual Colleen Cler Appeal

It has often been said that a Grandmother's love is limitless for her grandchildren. Char Ryan is no exception. For the last 10 years she has been writing a heartfelt appeal letter and sending it out to her family and friends in support of her granddaughter, Colleen who has Rett syndrome. Year after year she asks her loved ones for donations to enable Rettsyndrome.org to continue funding research that will one day lead to treatments and habilitative therapies that will make Colleen's future brighter. Through the generous donations that these letters have generated Char has been able to raise almost \$100,000. Special thanks to the Ryan and Cler families and all those who have supported this appeal through the years.

### Alex: For the Love of Her Sister

Everyday boys and girls from around the world raise their voices in support of their siblings with Rett syndrome. Alex is one example as she supports her younger sister, Sarah. The Varon family started their own foundation - the Sarah Varon Foundation to help raise awareness and funds for Rett syndrome research.

Alex began helping her sister at an early age. In order to communicate with her sister, Alex helped Sarah by learning the Minspeak language symbols and core vocabulary. She also helped her sister operate her speech generating device and came with Sarah on her weekly speech therapy sessions.

Alex and Sarah enjoy spending time together and even attend the same summer camp each year. They have been going to The Bridge School sleep away camp since 2006.



She has not only been an advocate for Sarah but for all children with Rett syndrome. Alex volunteered at the National Institute of Health study for Rett syndrome selling items to benefit the hospital and Rett syndrome research and watching younger siblings of the girls in the study. She also supported Rettsyndrome.org by hosting Lemonade Stands in front of her house and putting out an appeal through Instagram two summers in a row.



*Alex and Sarah*

We thank Alex, her parents Christine and Sol Varon and their family and friends who have generously supported Rettsyndrome.org through the Sarah Varon Foundation for the past 14 years!



*Craft a Cure for Keira*

## Bathie Family Shows Their Support

Jim and Katie Bathie are hosting their 2nd Annual Craft a Cure for Keira event July 15th in Milwaukee. The family started this event in 2015 as a way to gather their friends, family and community together in support of their daughter Keira. All money raised at the event goes to [Rettsyndrome.org](http://Rettsyndrome.org).

The Bathies are no strangers to fundraising. Starting in 2011 they hosted an annual Golf Tournament. Last year was the last year for the tournament as instead they wanted to focus their efforts on the Craft a Cure event.

Since they started hosting events in 2011, the Bathie Family has raised over \$235,000. We thank Jim and Katie for their commitment to fundraising to help find a cure for Rett syndrome.



*Jason Richards with daughter Camille and other Rett Racers.*

## Running the Distance for Rett Syndrome

Running has swept the nation as a popular way to exercise and have fun. [Rettsyndrome.org](http://Rettsyndrome.org) initiated a Rett Racer program several years ago to encourage those who love running to raise money for Rett syndrome in the process.

Jason Richards joined the Rett Racer program and has run many races in honor of his daughter, Camille. Recent races he has run include the Carlsbad Marathon, CA, America's Finest Race and the SoCal Ragner Race.

From his running pursuits he has raised over \$13,000 for [Rettsyndrome.org](http://Rettsyndrome.org). We thank Jason for using his passion to help raise money for his daughter and all those with Rett syndrome who can't run themselves!



**Join the Rett Racers** [Rettsyndrome.org](http://Rettsyndrome.org) has recently revamped our Rett Racers program. Check out our new, fresh website to learn more ([www.retracers.org](http://www.retracers.org)). Rett Racers include runners, cyclists, triathletes and more. Join as a Rett Racer for any existing race you are taking part in or work with us to create your own event! No fundraising minimums required. Thanks to all our Rett Racers for your efforts this year which have already brought in thousands of dollars that will be put towards research!

# Strolling and Rolling for Rett Syndrome

## An Interview with a Strollathon Chair

Putting on a Strollathon year after year takes dedication, hard work and the desire to do anything one can to raise funds for Rett syndrome. Rettsyndrome.org is honored to have many dedicated Strollathon chairs who have hosted events five, 10 and even 15 years in a row.



*Nebraska Strollathon*

We are honored to recognize Jacy Horst as one of our dedicated Strollathon chairs. This will be the 5th year that she has hosted the Nebraska Strollathon. She has grown the Stroll year after year despite many challenges. In 2015, she was able to obtain 23 corporate sponsorships; the most of any other Strollathon. We're excited to see what she has up her sleeve for 2016 and honored to recognize her for her efforts.

### *What is the most rewarding part of hosting a Strollathon?*

*The most rewarding part of the strollathon is giving the girls a day for THEM where THEY are the focus of love and support and not doctor's stuff! Raising awareness and money for our girls is a bonus too!!*

### *Where do you see the Stroll in the next five years?*

*In five years, I would like to see the stroll move around the state to continue to raise awareness across the state as well as tap into funds we are missing out on. I would like to see it west at least as far as Kearney and east to Lincoln and Omaha (all college towns!)....or anywhere else someone might be willing to go with it! A lot of companies don't want to donate if the walk isn't in that particular community. So by placing our stroll in some of these bigger college communities we may raise more money, get more volunteers, and of course, raise more awareness!*

## Updates to the Strollathon Program



*Starting Line*

In an effort to revamp Rettsyndrome.org's signature fundraising event the Strollathon program has seen some updates for the 2016 season. All of these changes have been spearheaded by Jackie Piscatelli, Family Development Manager for Rettsyndrome.org who has been charged with leading the Strollathons. Jackie has hit the ground running and made some exciting updates to the program.

- The Strollathon website ([www.strollathon.org](http://www.strollathon.org)) has undergone some dramatic changes making it more user friendly. Now when you log onto the site, it is clearly marked where you should click to be taken to the information you need. Also, a map shows users which states host Strollathons.
- A more streamlined process has been put in place to ensure that the role of Strollathon chair is as effortless as possible. Important resources have been placed at their fingertips to allow for easy access and use when soliciting donors, finding sponsors, organizing volunteers and generating excitement for their event.
- We want to continually add new cities and towns as locations to host Strollathons. This year we are excited to welcome six new Strollathons to the lineup. These include: Massachusetts, Idaho, Sulphur Springs, TX, Knoxville, TN & Clarksville, TN. We hope to add even more to this lineup for 2016.

# POSSIBILITIES

Each month we feature a child accomplishing and doing amazing things from participating in a Spartan Race to riding bikes. Just because our children have the diagnosis of Rett syndrome, it does not mean they cannot do everything their siblings and peers do. With a little effort and planning ahead as well as some patience and flexibility, your child can take part in a wide array of activities while remaining safe and sound. Participating feels good for the whole family but most importantly, it builds self-esteem.

In April we featured Bella for our Possibilities of the Month. Bella hadn't walked in a while, but after a lot of hard work and perseverance she got back to walking! The video showed Bella walking on her own! Check out the full video on our Facebook page - [facebook.com/rettsyndrome](https://facebook.com/rettsyndrome).

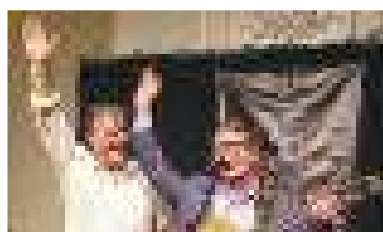


*Bella walking*

## Raising A Hand For Rett

On Tuesday night April 26, over 120 people gathered in Cincinnati to hear Kevin Black tell his story and perform a few songs. It was the first stop on a three city tour that Kevin and Dave Clements, co-creator and photographer of the book "Raising A Hand", were on to raise awareness and funds for Rett research. The Cincinnati event raised over \$50,000 and when combined with funds from prior book sales, Kevin and Dave were able to present a check to [Rettsyndrome.org](http://Rettsyndrome.org) for \$61,275!

Kevin and Dave are agreeable to doing a number of coordinated multi-site events to promote "Raising A Hand" and Rett awareness. At our June Conference books will be available for sale and we will have a number of times that fit into Kevin and Dave's schedule for any families that would like to speak with them about hosting an event and raising funds for Rett research. Kevin is an incredible entertainer who takes his audience on an emotional journey filled with laughter and tears, ending his performance with a not to be forgotten rendition of Lee Greenwoods, *God Bless The USA*.



*Kevin Black with committee member J.B. Buse*



*Kevin Black with Scott Farmer*

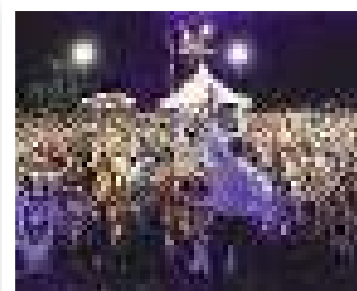
After leaving Cincinnati Kevin and Dave continued their journey north doing a show at Brimley, Michigan's Bay Mills Resort & Casino two nights later. Over 120 'locals' came out to support the cause and enjoy Kevin's music as well as Naomi Parish, who is featured in "Raising A Hand". She had her entire family there including grandmother Donna Wright who flew in from NY to be there. Also there were two other families with daughter's with RS and according to Dave "It sure looked like all three of the young ladies had a great time listening to Kevin sing."

The third stop on the tour had the guys in Hamilton, Ontario, Canada at the Bay City Music Hall doing a show and promoting the book in support of our north of the border Rett friends. As you know RS has no borders. This event was hosted by RS parents Darcy and Carrie Balak. All who were in attendance were appreciative of the effort and all 'raised their hands' high in support of rett research.

At the family conference we hope to tell you more about Dave and Kevin's book and their effort to help support the Foundation. On behalf of the entire Rett community we thank Kevin and Dave for the work they continue to do in support of [Rettsyndrome.org](http://Rettsyndrome.org).



*Kevin Black presents check to Anne and Gordy Rich.*



*On May 6th and 7th people Raised a Hand for Rett syndrome at the League City Music Festival in League City, TX.*



## 2016 Family Conference & Research Symposium

INFORMATION, CONNECTIVITY, COMMUNITY, EMPOWERMENT!



• JUNE 24-26, 2016 •

EAGLEWOOD RESORT - ITASCA, ILLINOIS  
RETT SYNDROME, MECP2 DUPLICATION,  
CDKL5 DISORDER & FOXP1 SYNDROME  
32ND ANNUAL FAMILY CONFERENCE

Don't miss the largest and best conference  
for all things Rett syndrome.

### PROGRAM HIGHLIGHTS NEW THIS YEAR:

- Family Research Readiness Arena
- Clinical trials, Natural History Study, Updates from the Labs
- Wearables and Monitors - Are They There Yet?
- Offsites to nearby ABILITIES EXPO
- Expand Horizons with our Related Disorders  
MECP2 Duplications, CDKL5 Disorder and  
FOXP1 Syndrome

SESSION TOPICS: Genetics, Medical and Symptom Management, Therapies, Communication and Literacy, Hi-Tech Eye Gaze How-To's for All Ages, School and IEPs, Adulthood Quality of Life, Fundraising, Advocacy, Awareness

**Register Now!** Online Registration Closes June 17th.  
[www.rettsyndrome.org/for-families/family-conference](http://www.rettsyndrome.org/for-families/family-conference)

**UNITY STRENGTH HOPE**

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

RESEARCHERS: Register by June 17

[rettsyndrome.org/research-programs/rett-syndrome-symposium-2016](http://rettsyndrome.org/research-programs/rett-syndrome-symposium-2016)

## What's In YOUR State?

### Important State Resources for Families

You may have noticed that the State Resources section of our website has a new look! We have several states that have been reformatted and researched and are ready to view. The new site also shows locations of clinics participating in the Natural History Study so parents can easily find those sites. Please take a look and let us know what you think.

As part of this project, we are recruiting parent volunteers to serve as State Resource Reps. This role is critical to the success and functionality of our state pages. As a State Resource Rep, you will connect families affected by Rett syndrome to information, tools, and resources. You will help families find local services and assist them in utilizing the State Resources page. The responsibilities of the State Resource Rep are extremely important and equally simple.

If this sounds like a role you would like to take on, email Project Manager Andrea Reed your name, phone number and state you would like to represent to [areed@rettsyndrome.org](mailto:areed@rettsyndrome.org) and she will contact you with more information.



## The 50th Anniversary of the First Publication on Rett Syndrome

It has been 50 years since Dr. Andreas Rett, an Austrian pediatric neurologist, originally described Rett syndrome in German medical literature. The 1966 article printed in German described Dr. Rett's observation of eight girls who bore similar clinical presentations and the hallmark trait of Rett syndrome—repetitive hand-washing movements or hand wringing. This first paper went largely unnoticed until Dr. Bengt Hagberg revealed this unique syndrome he named after Andreas Rett to an international medical meeting. Rett A. Über ein eigenartiges hirnatrophisches Syndrom bei Hyperammonämie im Kindesalter. Wien Med Wochenschr. 1966;116:723–728.

Rett syndrome research and patient care has come a long way since Dr.



Rett's initial observation of two girls in his waiting room sitting on their mother's laps presenting the same hand movements. Today, we have increased the quality of life for these girls and boys and their families affected by Rett syndrome by finding treatments, therapies, and best practices for care. We are excited for

additional treatments and a potential cure that will come through the drug development platform in the future and for the neuro-habilitation therapies that are key to development and maintenance of neural networks. We believe Dr. Rett would be proud of all the accomplishments that have been made for a neurodevelopmental disorder that was unknown 50 years ago.

To celebrate this hallmark anniversary, the international conference RTT 50.1 will be held from September 15 to 17 in the historic city of Vienna, home to Andreas Rett and the "birthplace" of Rett syndrome. RTT 50.1 is open to patients, clinicians, scientists, researchers and other healthcare professionals. For more information, please visit [www.rett2016.wien](http://www.rett2016.wien)

## Accelerating Research through the Scout Program with ANAVEX 2-73

After an NIH workshop that set the priorities for preclinical testing, Rettsyndrome.org forged ahead to design the Scout Program and proactively direct the research with PsychoGenics, a contract research organization that specializes in preclinical services and drug discovery. Aimed to accelerate research for potential treatments of Rett syndrome, the Scout Program is a battery of robust tests to check whether potential compounds or drugs can be used to reverse symptoms of Rett syndrome in the animal mouse model. The Scout Program is innovative in the way it attracts partnerships with academics, and biotech and the pharmaceutical industries. By providing a rigorous testing platform for their compounds, the program encourages collaboration with these groups to test either new or repurposed compounds that they believe might be useful in treating Rett syndrome without any risk.

This past spring, we have collaborated with Anavex Life Science Corp, a clinical-stage biopharmaceutical company to test their compound ANAVEX 2-73. The data generated by the Scout Program is owned by the company. In February, Anavex announced positive data of ANAVEX 2-73 in the Rett syndrome mouse model, and the company is now exploring the potential to advance ANAVEX 2-37 into human clinical trials for the treatment of Rett syndrome.

The data will be presented at the 14th Rett Syndrome Research Symposium by Christopher U. Missling, PhD, President and Chief Executive Officer of Anavex.

# *An appeal from our Chief Operating Officer*

## *Dear Friends,*

It is truly the time of Rett. 50 years ago Dr. Andreas Rett published his landmark paper describing eight girls who bore similar clinical presentations including the hallmark trait of Rett syndrome-repetitive hand washing movements or hand wringing. While much has been done in the ensuing 50 years, including reversing Rett symptoms in mouse models, the last couple of years have seen major strides being made in improving the quality of life for our girls. Rettsyndrome.org's strategy is now paying dividends as our research is yielding results in the here and now! From Clinical trials to neuro-habilitation studies, Rettsyndrome.org is funding research with results. Just look inside this newsletter at our best in class full spectrum research strategy and you'll see how your dollars have been put to work, and what a difference they have made. Research with Results, you deserve nothing less.

We have also made great strides in Family Empowerment, with our new web based State Resource Guide. Through Information-Education-Connectivity, Rettsyndrome.org is empowering families to be their daughter's strongest advocates. From our website to our Family Conference, to our Rett parents on staff and legion of volunteers, Rettsyndrome.org is here for the Rett community, any way you choose to access us.

None of this would be possible without a committed Board, generous donors, volunteers and staff. Under the guidance of Chairman of the Board, Rajat Shah, father to Sejal RTT, Rettsyndrome.org passed the \$38 million dollar mark for research investments that are making a difference today. Please take time to read his farewell letter, there is no way we can thank him for all he has done. As his multiple terms as Chairman of the Board come to an end this June, Rettsyndrome.org and the Rett community are in a better place due to his stewardship.

There is still so much we must accomplish. We have moved the ball from our own end-zone into the red-zone, and our goal of treatments and a cure are in sight. Moving more compounds into clinical trials and funding additional neuro-habilitation studies are primary areas of focus, but they are costly. Please know that the funds you have entrusted with us have been put to good use.

We are delivering on our promise of 'treatments' that will improve the quality of our girl's lives TODAY as we continue our search for the ultimate cure. But we need your help. Please join me in moving the ball across the goal line by donating today at Rettsyndrome.org or with the enclosed envelope.

**YOU CAN MAKE A DIFFERENCE!!**

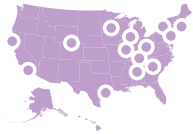
*Gordy Rich – Kelly, RTT C965T*  
Chief Operating Officer



*Gordy Rich and Family*



## Our Research Strategy ⇒ Best in class



**Three (3) multi-site clinical studies** advancing safe and effective pharmacological interventions, more in the pipeline



Pharmacological companies **seek out our SCOUT program** to test, and fail fast or succeed in moving more potential drugs into real treatments



**The only** Co-sponsor and Administrative coordinator of the 14-site NIH Natural History Study enrolling over 1,200



**Funder and trusted steward** of the Rett syndrome tissue research bank at Harvard which is open to ALL Rett researchers



Rettsyndrome.org is funding international databases, RettBASE and InterRett, that have **OPEN-ACCESS** to all researchers



**Only clinical and basic science** fellowship training program in the United States specifically designed for training tomorrow's Rett syndrome researchers



**The only** foundation concerned with establishing Neuro-Habilitation standards

## Full Spectrum Strategy ⇒ Research towards cure



Our strategy yields results in the here & now as we continue to fund and drive fundamental research towards a cure, and foster next-gen researchers and clinicians through our Fellowship program.

# We are leading the revolution in Rett syndrome research

50 years later, Dr. Andreas Rett (est. 1966) would be proud



# We are results driven for here & now improvements in quality of life for all with Rett syndrome

# The Path to Drug Development



ENTREPRENEURSHIP  
WITHIN RESEARCH

**Fundamental Science or Basic Research**  
Centers about scientific inquiry  
Time: Unknown

Typical grant for Basic and Translational Research  
Centers for SCARF Library of compounds to screen available  
for researchers

Monopos or David Drenth's  
helps to research



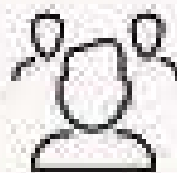
**Pre-clinical development**  
Along road of  
clinical trial

Conducts basic and pre-clinical  
studies through the  
Animal Program

Testing compounds  
before  
Pre-clinical  
Research

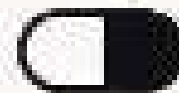
Moves with pharmaceuticals from  
companies and the FDA during the  
planning stages of clinical trials

Plays an integral  
role in the  
pre-clinical  
development of  
clinical research



**Clinical Trials and Research**  
The drug  
is being  
tested

Approved to  
FDA  
Time: 12 years



The process of creating new medicines is complex, time-consuming and costly. Moving a potential therapy from concept to being FDA-approved takes between 10 and 15 years and cost development of much more than \$1 billion.

Together, we can make this journey to the future.

ENTREPRENEURSHIP WITHIN RESEARCH  [RettSyndrome.org](http://RettSyndrome.org)

## Empowerment ➞ Through Knowledge



Rett parents on staff as well as filling volunteer positions, ready to help



A comprehensive, information packed website with a state by state resource guide



Annual Family Conference bringing the best, most relevant resources together



Support and management of Special Interest Networks as well as parent email Listserv- The RettNet



Full time Family Development Manager to help families with their fundraising efforts

✓ Information

✓ Education

✓ Connectivity

**Donate** ➞ [www.rettsyndrome.org/donate](http://www.rettsyndrome.org/donate)

**Participate** ➞ [www.rettsyndrome.org/get-involved](http://www.rettsyndrome.org/get-involved)

**Sign up** ➞ [www.rettsyndrome.org/join](http://www.rettsyndrome.org/join)



Rettsyndrome.org is a 501c(3) non-profit corporation registered as The International Rett Syndrome Foundation and established in 2007 through the strategic merger of the Rett Syndrome Research Foundation and the International Rett Syndrome Association.



538M 



We are leading the revolution  
in Rett syndrome research.

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Journal of Management Inquiry 22(2)

**195**  **CD**  
 (1995) CASSELL'S BOOKS/STRENGTH OF CHARACTER



★ ★ ★ ★  
**STAR CHARITY**  
 YOUNG BARRISTERS CHARITY  
 FUNDRAISING SOCIETY  
 2012-2013



We invested \$2.1 million to support new initiatives, grants, internships and research programs, and created a new Health-Driven Program.

Dehydration was administered 3 additional times to the WH Normal Military Study, which was run over a total of 35 participating sites. This separate effort, however, was initiated 10 days across the season, for our first 4 weeks.

In total, we have described over 225 published and unpublished research studies in a range of ecological, theoretical, and field topics.

Deaflympians are using a petition to persuade the British Government to undertake an official advocacy campaign in January 2015 to enable deaflympics research to be funded by the Department of Health. The campaign was a success and now deaflympics research is directly in scope of fully eligible for costs SRTD review of GdC research funds.

Test Number  
0042-2565

- Phase 2 trial in adult women closed in September 2016
- Moving forward with a Phase 2 trial for safety and tolerability in children with RDS, cystinosis
- Concurrently, Nappes Pharmaceuticals is in discussion with the FDA on design of Phase 1 pediatric study with 10 mg/kg as a treatment of RDS in neonates

## WF 1

- After meeting with the FOM they recommended the expansion of the SC-1 trial to include 17 additional patients.
- Added: Chapter 11, and 6 new cases, SC as a site.

# IMPACT REPORT

2015



## OUR MISSION



Accelerate full spectrum research for treatments and a cure for Rett syndrome



Empower families with knowledge and connectivity

2016

## LOOKING AHEAD

- ➔ Launch of new Phase 2 clinical trial with trofinetide in a pediatric population with Rett syndrome
- ➔ Expansion of the IGF-I human clinical trial in Boston, MA, Chicago, IL and Greenwood, SC
- ➔ Enrollments to the NHS at 14 sites throughout the U.S.
- ➔ 2016 Rett Syndrome Research Symposium and first ever Related Disorders Family Conference



## DONATE

[www.rettsyndrome.org/donate](http://www.rettsyndrome.org/donate)



## PARTICIPATE

[www.rettsyndrome.org/get-involved](http://www.rettsyndrome.org/get-involved)



## SIGN UP

[www.rettsyndrome.org/join](http://www.rettsyndrome.org/join)



Rettsyndrome.org is a 501(c)(3) non-profit corporation registered as The International Rett Syndrome Foundation and established in 2007 through the strategic merger of the Rett Syndrome Research Foundation and the International Rett Syndrome Association

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🌐 <http://www.rettsyndrome.org/connect> | [f /rettsyndrome](https://www.facebook.com/rettsyndrome) | [@rettsyndrome](https://twitter.com/rettsyndrome)

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# *A letter from outgoing Chairman of the Board Rajat Shah*



Simran and Sejal Shah

My daughter Sejal turned 10 on April 22. She is growing up to be a beautiful young lady. She brings a smile to my face every morning, and I can't imagine life without her. Yet, at the same time, she also brings me the most unbearable heart-break.

Eight years have passed since Sejal's diagnosis with Rett syndrome. I recall that May afternoon when Jenny and I received the diagnosis - like it was yesterday. The tears running down the faces of the doctor and other staff when they delivered us the news. I remember, too, our follow up visit a few days later where the message was simply 'take her home and love her; there is nothing more you can do.'

Although well-meaning, that was not a message I could live with for my daughter. A few days later, I remember holding Sejal in my arms, telling her that I was a "fixer" and promised that I would cure her. I would give up everything I have, everything I am for Sejal to have a typical, normal life; for our daughter Simran to have the best friend we promised was on the way when Jenny was pregnant with Sejal; and to walk Sejal down the aisle one day. Although at times I feel like a failure because Sejal hasn't been cured, the truth is -we are succeeding in so many ways.

Today, our families are in a much better place than we were eight years ago. We have spent over \$3.3 million to place our girls in four human clinical trials with the potential of the first FDA-approved treatments for Rett within sight:

First IGF-1 trial (completed) at Boston Children's Hospital.

IGF-1 Trial (ongoing) at Boston Children's Hospital, Rush Medical Center in Chicago and Greenwood Genetic Center in South Carolina. First trofinetide trial (completed) was at Baylor College of Medicine in Houston, University of Alabama in Birmingham, and the Gillette Children's Specialty Healthcare in Minneapolis-St. Paul.

Second trofinetide study (ongoing) at UAB, BCM, Gillette, Greenwood, Boston Children's, UCSD, UCSF, Cincinnati Children's Hospital, Rush, Children's Hospital of Colorado, and Vanderbilt. See the following web page: [www.rettstudy.com](http://www.rettstudy.com)

The Scout Program has had two successes and both of these drug companies have applied for Orphan Drug Status and are planning future clinical trials. TRC/Revive with their compound Tianeptine and Anavex with their compound Anavex 2-73.

Although IGF-1 and trofinetide will not cure our girls, early reports indicate they will improve their quality of life.

How amazing will it be for our daughters to sleep through the night, to breathe, swallow and chew normally, to regain their ability to walk, to potentially say a few words again, to have less anxiety, and to have functional use of their hands? Any one of these changes will open up a world of possibilities for them.

With more funding, we will find more potential treatments and advance additional human clinical trials for compounds already in our pipeline. We created the Scout program three years ago to do just that. As a result, drug companies are actively seeking us out to test more drugs for potential treatments and a cure.

Those who know me know that I can be impatient at times. Rett syndrome and caring for Sejal have taught me to think in terms of a marathon and not a sprint. Although we all desperately want a cure, we still need to push daily to maximize our girls' quality of life. This truth led the Board of Rettsyndrome.org in 2014 to invest for the first time in research into therapies to enhance the present quality of life for our girls. The goal is simple: to fund research to ensure that therapeutic strategies are focused on the complexities our girls face to maximize their chance to walk, to effectively use their eye gaze devices to communicate, to improve upon purposeful use of their hands, and to better assess cognitive function so we can understand the girls' needs. Until we find that cure, our girls deserve therapy and services geared toward giving them the fullest and healthiest lives possible.

Rettsyndrome.org has invested nearly \$40 million in research. These funds have provided tangible results but this is not enough. Even if Rettsyndrome.org is able to fulfill its goal of funding \$10 million a year in research for each of the next 10 years, we are only scratching the surface. That is why I want to thank you for

helping us get much-needed federal help over the last two years. With our lobbying in Washington D.C. and your support through letters and phone calls, we were able to get the NIH to expand the 14-site Natural History Study which will enroll over 1,200 girls and women. Because of our joint efforts, Rettsyndrome.org is the only co-sponsor and the administrative coordinator of this initiative. Also, last year we worked with Congressional House and Senate members to pass a law designating Rett syndrome as one of only 40 areas of study eligible for up to \$278 million in federal funding under the Department of Defense's Peer-Reviewed Medical Research Program.

Although I haven't cured Sejal, for the first time in these eight years, I have real hope for the future. We are much closer today than ever before, and we are on the right path. With your continued support and commitment, I know we will get there.

We continue to look for committed individuals to carry on this fight for our girls – to give them back their voices. If you are interested in hosting or co-hosting a fundraiser, please contact Gordy Rich at [grich@rettsyndrome.org](mailto:grich@rettsyndrome.org) and he will be happy to help. We are also looking for individuals for our Board with a passion and drive for our girls and who are able to give or raise significant funds for our continued research. If you are interested and have the capacity, please contact Gordy, he would welcome the chance to talk with you!

Finally, to our Rett syndrome family, I hope to see you in Chicago in June for the Family Conference and Research Symposium. This will be my last family conference as Chairman of Rettsyndrome.org. I know I leave our organization in good hands and I believe the past three years have pushed our organization and community to a better place.

All best, always,  
Rajat



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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.*