



# Rett GAZETTE

Accelerating Research.  
Empowering Families.



## A DECADE OF RESULTS



Rettsyndrome.org

ACCELERATING RESEARCH. EMPOWERING FAMILIES.

Chief Science  
Officer Letter **p3**

## The Reality of Rett Research

by Gordy Rich, COO, Rettsyndrome.org

I am so excited about all of the recent good news surrounding Rett syndrome research. We have accomplished much in the past 10 years and even more is happening RIGHT NOW! We are seeing advances in pharmacologic, neurologic and genetic interventions that will transform lives. Please look at our new infographic on page six to see all we anticipate in the 'short term'. From drugs and biologics to neuro-habilitation advances, our research strategy is yielding actionable outcomes, not false promises.

With all the achievements, it is important to remember that there is still critical work to be done. When our daughter Kelly was diagnosed with Rett syndrome over 20 years ago, there was no known cause, treatment, or hope for a cure. That changed when mutations in the *MECP2* gene were discovered to be the cause of Rett syndrome. When we saw the Rett mouse model's symptoms turned off, we felt such hope. We also saw several potential treatments claiming to be the 'cure' for Rett syndrome fail to live up to their promise. From stem cells and bone marrow to gene therapy,



Gordy Rich

all have shown potential but none, to date, have proven safe enough to play a part in unlocking the complexities of Rett.

The reality of Rett research today is that there are a number of pharmacologic compounds in the clinical trial process; many more than what we had even five years ago. When these compounds become prescribable, they will help individuals with Rett syndrome of all ages. We believe that in the future, pharmacologic and genetic interventions will work in tandem to correct the biology of Rett syndrome. We also believe that the neurology must be reset to make up for missed developmental milestones. Unfortunately, this means no one-time fix for Rett syndrome. The good news is that researchers are making biologic and neurologic advances and Rettsyndrome.org is supporting them. We continue to seek out the most credible and expert research, with the highest probability of success, so that we can positively impact the lives of all of those with Rett syndrome. Please join us so that, **together, we can accelerate the pace of research and empower families with its results!**

# A Decade of Research – A Celebration of Many Advances

by Steve Kaminsky, PhD, Chief Science Officer



Steve Kaminsky, PhD



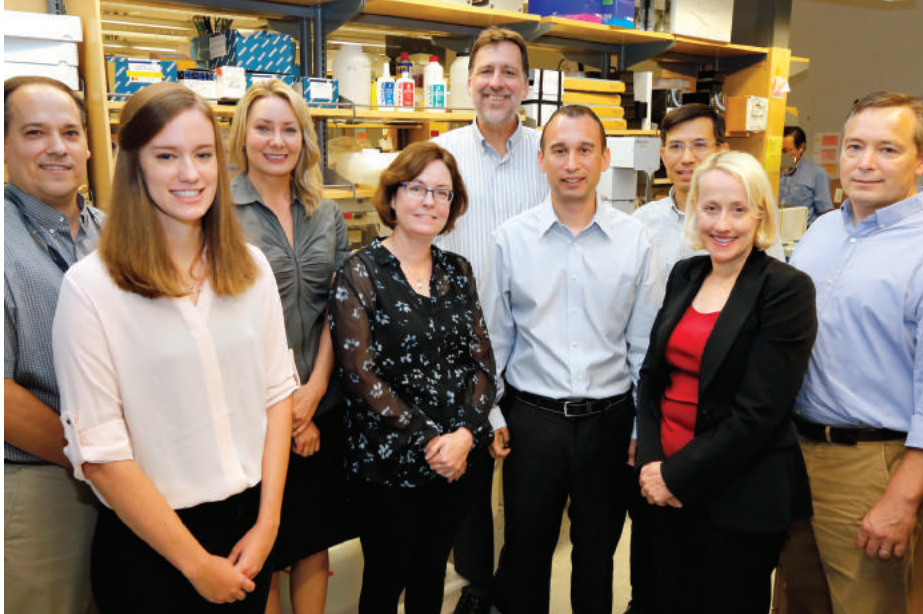
2007 marked the beginning of a momentous decade for Rett syndrome research, and I want to celebrate those achievements. Ten years ago, a hallmark scientific paper on Rett syndrome was published by Dr. Adrian Bird titled “Reversal of neurological defects in a mouse model of Rett syndrome”. His research was the first to demonstrate that genetic manipulation of the Rett syndrome mouse model led to improvements of the Rett-like symptoms in the mice. This study gave the community hope that Rett syndrome could be “reversed” if the *MECP2* gene could be normalized. That same year, four more papers were published showing that pharmaceutical interventions could also improve the Rett-like symptoms in mice. The research community started on the path to find treatments and a cure for Rett syndrome.

In this past decade, over two dozen research articles have been published showing that there are many different compounds that can improve Rett-like symptoms in mice. Rettsyndrome.org’s Research to Reality Program has been focused on the drugs and compounds outlined in

these articles, bringing those with robust proof of principle studies through translational research and into clinical trials to reach the bedside as approved medicines for Rett syndrome. We have seen success in our Scout Program testing potential drugs in the animal model. Our Scout Program allows us to rapidly identify compounds that will correct the biology associated with Rett syndrome efficiently and safely.

We have also seen some advancement in genetic interventions such as gene therapy. In this last decade, five papers have been published regarding gene therapy for Rett syndrome. We have written about the great progress with gene therapy, and the long road ahead to make it safe and effective.

Ten years of hard work have moved research much closer to reality. While we do not yet have an approved medicine, we are closer than ever before. We continue to accelerate research to correct Rett biology and re-build neurological networks. We look forward to another decade of advances that will improve the lives of all those suffering with Rett syndrome.



Steve Green, Vanderbilt University

## Advocate and Serve

“Advocate and serve. High-impact organizations don’t just focus on doing one thing well. They may start out providing great programs, but eventually they realize that they cannot achieve systemic change through service delivery alone. So they add policy advocacy to access government resources or to change legislation, thus expanding their impact.

*(Forces for Good, authors Leslie R. Crutchfield & Heather McLeod Grant)*

In 2015, Rettsyndrome.org drove a successful advocacy campaign that brought attention to Rett syndrome research, and Congress added Rett syndrome as a topic of interest to the research funding program called the Peer Reviewed Medical Research Program (PRMRP). The PRMRP supports research across the full range of science and medicine, with a goal of enhancing the health and

well-being of military Service members, veterans, retirees, and their family members. You may ask why the PRMRP would want Rett syndrome to be a topic of interest. Rett syndrome is a brain synaptic disorder, much like Traumatic Brain Injury that is common in the military. By studying Rett syndrome, the hope is to bring novel treatments for all of those suffering with brain disorders. With Congress’s

inclusion of Rett syndrome, our researcher community can apply for funds appropriated by the Senate (\$278M in 2016 and \$300M in 2017). We are pleased that our advocacy efforts allowed researchers

access to this pool of research dollars that is often difficult to obtain by any single non-profit organization. Our researchers submitted applications for funding and the PRMRP has announced the results of the 2016 grant cycle. Eight applications related to Rett syndrome were received. One application was funded at nearly **\$1.9M dollars.**

Needless to say, we are ecstatic to share the news that one of our previously funded grantees applied for and received a highly competitive three-year award from this DOD program. Colleen Niswender, PhD, Professor of Pharmacology and Director of Molecular Pharmacology at the Vanderbilt Center for Neuroscience Drug Discovery in Nashville, TN received a 2016 Investigator-Initiated Award for the project

“The Role of Metabotropic Glutamate 7 in the Etiology and Treatment of Rett Syndrome”. The Investigator-Initiated Research Award is designed to support research with the potential to yield highly impactful data that could lead to critical

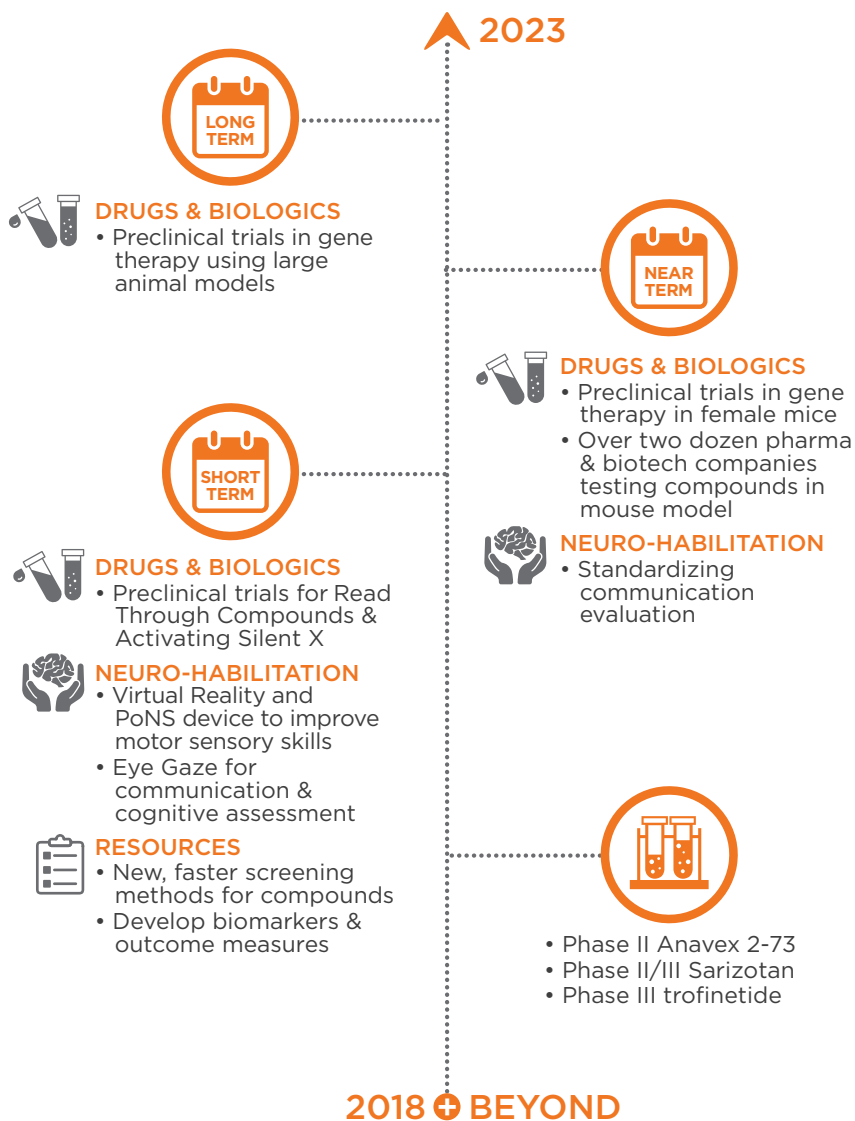
discoveries or major advancements. “We are very excited to receive support for this project from the PRMRP, which is managed by the Department of Defense’s Congressionally Directed Medical Research Programs (CDMRP),” said Dr. Niswender. “Our work on Rett syndrome began with a critical seed grant from

“**We are very excited to receive support for this project from the Department of Defense’s Congressionally Directed Medical Research Programs (CDMRP),” said Dr. Niswender. “Our work on Rett syndrome began with a critical seed grant from Rettsyndrome.org in 2012. We believe that our studies in Rett syndrome models will provide important information into therapeutic possibilities for both Rett syndrome patients as well as individuals with brain injuries such as those that affect military populations.**

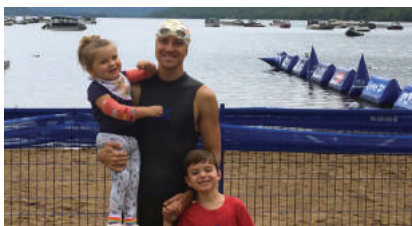
Rettsyndrome.org in 2012. We believe that our studies in Rett syndrome models will provide important information into therapeutic possibilities for both Rett syndrome patients as well as individuals with brain injuries such as those that affect military populations.” You can read Dr. Niswender’s abstract at [www.rettysyndrome.org/for-researchers/investigator-spotlight](http://www.rettysyndrome.org/for-researchers/investigator-spotlight).

# The Path Forward

During this last decade, Rett syndrome research has made tremendous progress. In the decade to come, we anticipate more treatments becoming available and more progress made on our path to a cure for Rett syndrome. **The best is yet to come.**



## Rett Racer Feature – Sean Doyle



This summer, Sean Doyle's daughter, Isla, was diagnosed with Rett syndrome. Sean used his passion for athletics to dive into fundraising. He signed up for the Ironman Mont-Tremblant in Quebec. The 2.4 mile swim, 112 mile bike ride and 26.2 mile run is no comparison to what Sean said his daughter Isla experiences. The support from

his friends, family and business was astounding. In just four weeks, Sean raised over \$20,000 for Rettsyndrome.org! Sean's employer, Time Warner, has been gracious enough to match a portion of funds raised. We are so grateful for the support of Sean, his community and Time Warner. Job well done Sean!



## Stroll Across the Country

This Spring and Fall, Strollathons took place in over 30 cities across the United States. These family-friendly events allowed families to connect not only with each other, but with their communities. From carnival themes to princesses, the fun never ended. Each Stroll brought us closer to our million dollar goal and we can't wait to see what 2018 brings.



## Casting 4 a Cure Reels it in Again

Casting 4 a Cure was another huge success this year. Fly-fishing enthusiasts from across the country gathered to fish and fundraise this past August. Over the course of 10 years, Casting 4 a Cure has been extremely supportive of Rettsyndrome.org and donated over \$600,000 dollars.

(800) 818-7388 | <http://www.rettsyndrome.org> | [/rettsyndrome](https://www.facebook.com/rettsyndrome)  
[/rettsyndrome](https://www.instagram.com/rettsyndrome) | [/youtube.com/irsfoundation](https://www.youtube.com/irsfoundation)

## Welcome Melissa Kennedy, Our New Executive Director



Rettsyndrome.org is pleased to welcome Melissa Kennedy as our new Executive Director. Melissa is a seasoned health care professional with a clear passion for our mission and strategy. With her help, Rettsyndrome.org can build bridges with organizations that share our mission to treat and cure neurological diseases such as Autism and Parkinson's. These partnerships will allow us to accomplish more in less time. Melissa, along with the dedicated staff of Rettsyndrome.org,

will continue to empower our community.

Melissa received her Master in Health Administration from The Ohio State University. She co-founded a healthcare research company and later worked at different medical institutions, including TriHealth, developing strategy and directing new programs.

Melissa lives in Wyoming, Ohio, with her husband John and her two sons, Patrick and Will.



**A DECADE OF RESULTS:  
THE BEST IS YET TO COME**

Get more in-depth information on our research  
by visiting [rettsyndrome.org/decadeofresults](http://rettsyndrome.org/decadeofresults)