RSE Quarterly E-News - February 2025

Upcoming Events!

European Congress in Rett Syndrome 2025 – 11th - 12th October 2025

The 2025 Rett Syndrome European Congress will be held in Istanbul, Turkey on 11th - 12th October 2025.

The conference is being organised by the Turkish Rett Syndrome Association under the leadership of RSE.

The slogan of the conference is "**The Convergence of Science and Hope in Rett Syndrome**" due to the launch of DayBue as the first commercial drug for Rett syndrome in 2023 and the ongoing process of genetic treatment studies evolving into human trials.

In accordance with the slogan, the main themes of the conference were determined under the following headings.

1. The Convergence of Science and Hope in Rett Syndrome
2. Rett Syndrome: The Path to Genetic Treatment
3. Shaping the Future: Groundbreaking Solutions in Rett Syndrome
4. Rett Syndrome with International Perspectives

Purpose of the conference: This conference aims to raise awareness in society while addressing developments and scientific collaborations in the treatment of Rett syndrome. We aim to share information on a wide range of topics from genetic research to clinical applications.

At the end of the conference, we will draw attention to the power of international collaboration to improve the quality of life of individuals with Rett syndrome.

European Congress in Rett Syndrome 2025 – 11th-12th October 2025

The content of the conference offers a broad perspective from current scientific studies to clinical applications. Each day, there will be an opportunity to learn about Rett syndrome from a different perspective.

Featured topics:

1. Genetic research, neurology, speech therapy, physiotherapy, nutrition, gynaecology, and drug applications.
2. Global cooperation and international awareness studies.

Shaping the Future Together

This conference is a meeting point for scientists, medical professionals, families and volunteers. Our goal is to research and share new treatment methods that will improve the quality of life of individuals with Rett syndrome.

Your participation will help draw attention to these important issues and raise global awareness.

Rare Disease Day February 28th 2025

Rare Disease Day Official Video here – How can you get involved? Share your story here - https://www.rarediseaseday.org/share-your-story/

You can also use their social media resources to bring attention to the campaign https://www.rarediseaseday.org/downloads/ These are available in multiple languages. All free to download!

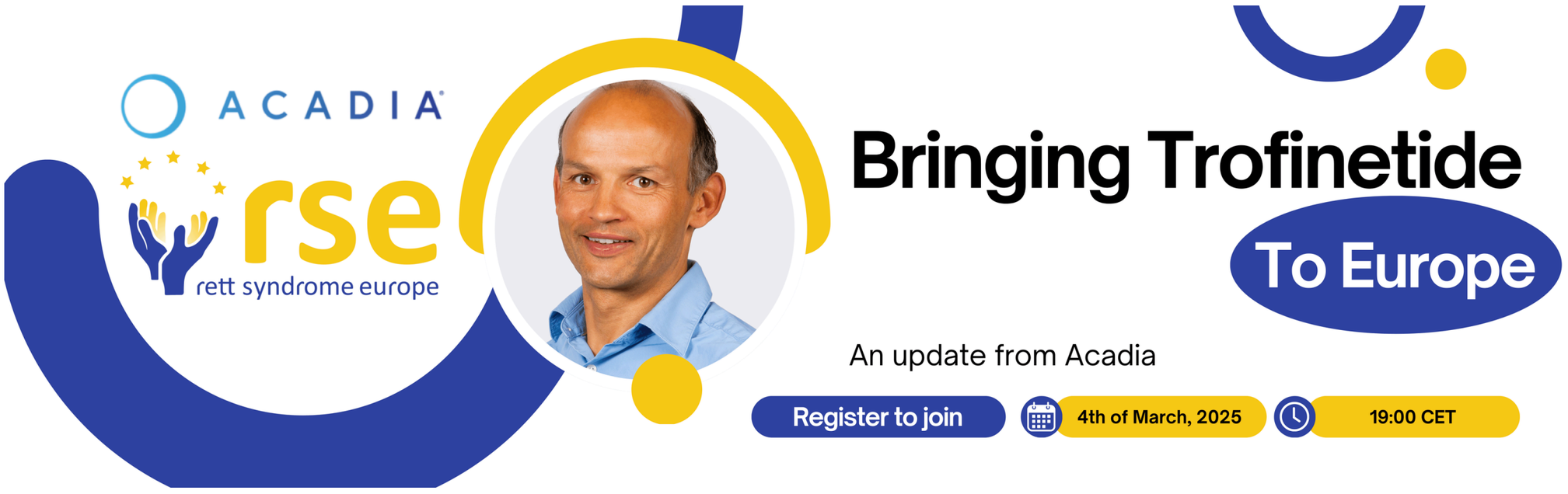
To mark **Rare Diseases Day 2025**, board member Burak Temiz will be a panelist/speaker at the panel organised by Istanbul Undiagnosed and Rare Diseases Platform (www.istisna.org) on February 25th 2025.

RSE Six Monthly Membership Meeting

RSE Six Monthly Membership Meeting – we are looking at some dates in May 25 to hold this and have created this poll for you to complete. Apologies as it was set to go out before Christmas but for some reason got stuck in the outbox and has only just been released to your inboxes! Thanks to everyone who has completed it so far but if you have not yet please do so asap. Here is the link https://doodle.com/meeting/participate/id/dGz5OR3b at the moment Wednesday 7th May 6-7pm CET looks like the most popular date and time.

Please submit your votes before the end of February so we can confirm the date!

Meeting with Acadia about their application to EMA



Great opportunity to hear from Tom Pulles and Stephanie Kim at Acadia about their application to the EMA for approval of trofinetide and also the care giver research they are undertaking to support the application. Save the date 4th March 2025, 7pm CET Here is the meeting link. Hope you can join us!

Research

The gut issues in Rett syndrome – more common and problematic than perhaps doctors think? This recent study highlights the different ways the gut is affected and just how common a problem it is with over 80% of individuals affected. Doctors though may not be aware that this is the true extent of the problem so more awareness raising is needed in this area. https://rettsyndromenews.com/news/gi-symptoms-affect-80-rett-patients-study/

We are also reading more and more about the importance of a healthy gut microbiome and how this can be supported in someone with Rett so this will be a topic in this year's congress in Istanbul.

New study from America here that offers hope for safer and more targeted gene therapies as well as insights into earlier molecular changes in the brain particular in the hippocampus. https://www.drugtargetreview.com/news/155364/rett-syndrome-discovery-could-lead-to-better-treatments/

"We discovered several candidate biomarkers sensitive to MECP2 levels that could be the key to developing safe gene therapies for Rett," he said. "Our study more broadly demonstrates the importance of cataloguing and understanding the earliest biological events that occur during symptom onset in neurodevelopmental disorders."

Update from Anavex

Anavex Lifesciences has completed data collection for the Phase 2/3 EXCELLENCE Clinical Study in Pediatric Rett Syndrome at sites in Canada, Australia, and the UK. Over 91% of the patients completing the trial continued on into a 48-week open-label extension study

A Compassionate Use Program has also been established in these countries to ensure access to patient participants, and 93% of those who completed the open-label extension have joined the programme. There is a deep appreciation to all of the families who participated in this first study of ANAVEX®2-73 in pediatric patients with Rett syndrome. Please see https://www.anavex.com/post/anavex-2-73-receives-compassionate-use-authorization-for-pediatric-patients-with-rett-syndrome

Research

Daniela Tropea, a researcher in Dublin, Ireland is looking for funding support for a PHD Research Student Project – This is the abstract below for the project:

Rett Syndrome (RTT) is a rare condition that's one of the main reasons for disabilities among girls. At first, everything seems normal with the kids, but when they get to around 1 or 2 years old, they start showing signs of the illness. They might have trouble moving or talking, find it hard to breathe, do the same things with their hands over and over, or have seizures. Right now, there isn’t a cure for RTT, so doctors focus on taking care of these symptoms, and many people with RTT need help throughout their lives. Most of the time, RTT is linked to a change in a specific gene, but the symptoms can be very different from person to person, which makes it hard to predict and treat correctly. In research, patients often react differently to treatments, which makes scientists think there are various types within RTT. We haven't figured out a way to identify these types yet, but looking at how the brain functions might help us to do that.
This proposal has three aims:

AIM 1: Gather demographic, molecular, and EEG information from RTT patients and comparable controls.
AIM 2: Use quantitative EEG to analyse brain activity and discern unique EEG patterns that can guide diagnosis and predict disease progression.

AIM 3: Boost awareness of RTT cases by integrating EEG data into DATARETT- a data sharing platform created to collect and share research data.

The outcome of this research is threefold:

1. it will improve patients’ prognosis and management,
2. it will improve the understanding of RTT biology, and 3) it will enhance the visibility of patients with RTT.

Link to call - https://www.researchireland.ie/funding/enterprise-partnership-postgraduate/

The funding partner/s would be expected to contribute 11,300 Euros per annum for 3 years to the project. This could be several patient organisations making a contribution but also if there was an interested company that is eligible too as a partner.

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Please contact daniela.tropea@tcd.ie for more information.

Other News

We're excited to share that we're in the process of setting up rettX, our new European patient registry for Rett syndrome. Although it's still being implemented, this registry is designed to bring together families, doctors, and researchers to learn more about Rett syndrome and find better ways to care for those affected. By collecting important information on everything from clinical care to personal experiences, the registry will help guide future research and treatments.

We believe that getting the community involved early on is key. As we continue to build and improve the registry, your participation and feedback will be invaluable. Whether you're a family member, a healthcare provider, or a researcher, your input can help shape a resource that makes a real difference in understanding and managing Rett syndrome. Stay tuned for more updates as we roll out this exciting project!

Advancing Genetic Medicines via the Rett Syndrome Global Registry

by Jana von Hehn, PhD, Chief Scientific Officer, Head of Clinical Development, RSRT

Imagine the wealth of information every Rett syndrome parent has about their child. Collectively, that data is critical to pharmaceutical companies developing medicines for Rett but until now, was not available to them.

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The Rett Syndrome Global Registry is an international database created by the Rett Syndrome Research Trust to gather standardized, information about individuals with Rett syndrome in an effort to accelerate genetic medicines and drug development.

Other News

From the **comfort of hom**e and available in **10 languages,** parents provide information on medical history, therapies, education, quality of life, and more, creating a comprehensive overview of how Rett is experienced and managed across the globe.

The goals of the Rett Global Registry include:

1. Accelerate the development and commercialization of genetic medicines and drugs for Rett by providing researchers access to a large pool of anonymized patient data.
2. Incentivize more biopharma to pursue Rett as a disease indication
3. Improve patient care
4. Identify and fill gaps in care
5. The use of registry data in lieu of a placebo control
6. Identify patients eligible for clinical trials
7. Amplify family voices about your child's Rett journey

Importantly, data from the Rett Global Registry will be **shared back with parents**. We are currently preparing the first Annual Report and look forward to sharing it with the global Rett community soon.

There is **power in numbers**. Every person that joins makes the dataset more accurate and more robust. Increasing participation in the registry by European families ensures faster access to treatments both during development and once approved. Participation in the Rett Global Registry does not limit participation in other registries or research initiatives.

To read more about the Rett Global Registry click here.

We encourage you to join today!

Family Focus

Two inspiring and heartwarming Family Focus stories for you to read. Many thanks to both families for taking the time to write this for us and share something of their lives with Rett syndrome. It always helps others on this journey to know you are not alone in your struggles and there is so much joy to be found still.

Our daughter Niki was born in 2007. "Niki" (Νίκη in Greek) is translated in English as "victory". Before she was born, I was trying to live my life under my moral compass, always do the right thing. I was thinking, If not trying to do the right thing then nothing in this universe has any meaning. That was my life motto at that time.
When we had the first signs of Niki's regression, I thought to myself, what is this? Is this a punishment? A reward? Is this a lesson? A lesson for who? And then she was diagnosed with Rett syndrome

Niki Konstaninou Without Wings

To be perfectly honest, more than once this past spring, I found myself asking: “Is this madness? Should we even dare? Are we even allowed to have this life?”
We were facing a packed itinerary for the summer—Germany, Poland, Italy, and India—bringing equal parts trepidation and excitement. What started as travel surrounding a study abroad program I developed for the University of Georgia, entitled Language, Power, and Globalization, set in Munich and Berlin, quickly expanded. Invitations for academic talks on my research with my daughter, Kalika, who is eight years old and has Rett syndrome, arrived from Poland and Italy. And since we would be already halfway there, we added a visit to my hometown of New Delhi, India.

It wasn’t a decision we took lightly. Months of preparation went into this—planning medication schedules, packing for different climates, coordinating medical equipment, and what seemed to be a million other things. And of course, making sure our cats would be cared for back home.

Adventures in Accessibility: Exploring the World with Rett Syndrome