



Network Support & Research Event March 2023

Becky Jenner, RSE President



Aims of the event

To improve the quality of life for people with Rett syndrome and their families in countries where support, information and advice is lacking particularly amongst health care professionals, medical specialists and educators.

To encourage new research collaborations particularly in areas where there are gaps in knowledge.

To aid the development of patient organisations to facilitate clinical trial recruitment and engagement with EMA and regulatory bodies for new and emerging treatments including gene therapy.

Main meeting in Hungary

- Satellite meetings in UK, Netherlands, Croatia, Georgia, Turkey and Serbia.
- 52 attendees in person in Hungary.
- 36 in Turkey
- 19 in Serbia
- 11 in Georgia
- 21 talks
- 6 languages translated using Veedio and parents help!



Day 1 Friday 17th March



Network Support and Research Meeting 17th & 18th March 2023



09:30 - 09:45	Introduction	Becky Jenner & Gillian Townend
09:45 - 10:15	Rett Syndrome: The Road from Recognition to Treatment	Jeffrey Neul
10:15 - 10:35	Tea Break	
10:35 - 10:50	Gene Therapy: Hope, Myth and Difficulty	Jean-Christophe Roux
10:50 - 11:05	Human Stem Cell-based Models for the Study of Rett Syndrome: Overview and Perspectives	Sonja Guil
11:05 - 11:35	Discussion in Country Groups	
11:35 - 12:00	Q&A to Speaker Panel	
12:00 - 13:15	Lunch / Networking	
13:15 - 13:35	Genetic Therapy Approaches in Rett Syndrome - an update	Stuart Cobb
13:35 - 13:45	Neurogene Update: NGN-401: A Self-regulating Gene Therapy for Rett Syndrome	Stuart Cobb
13:45 - 14:00	Neuren Update: Trofinetide in Rett Syndrome	Nancy Jones
14:00 - 14:10	Taysha Update: An Investigational Approach to Gene Therapy for Rett Syndrome	Benit Maru
14:10 - 14:20	Burden of Illness in Rett Syndrome: Initial Evaluation of a Disorder-Specific Caregiver Survey	Walter Kaufmann
14:20 - 14:35	Clinical Trial Registries and Databases	Daniela Tropea
14:35 - 14:55	Patients at the European Medicines Agency	Francois Houyez
14:55 - 15:15	Tea Break	
15:15 - 15:25	Family Contact and Engagement: Why and How	Becky Jenner
15:25 - 16:00	Discussion in Country Groups	
16:00 - 17:00	Q&A to Speaker Panel and Whole Group Discussion	
17:00 - 18:30	Drinks / Networking / Chat *	
18:30 - 21:00	Dinner *	

* organised for people at the in-person events



Day 2 Saturday 18th March



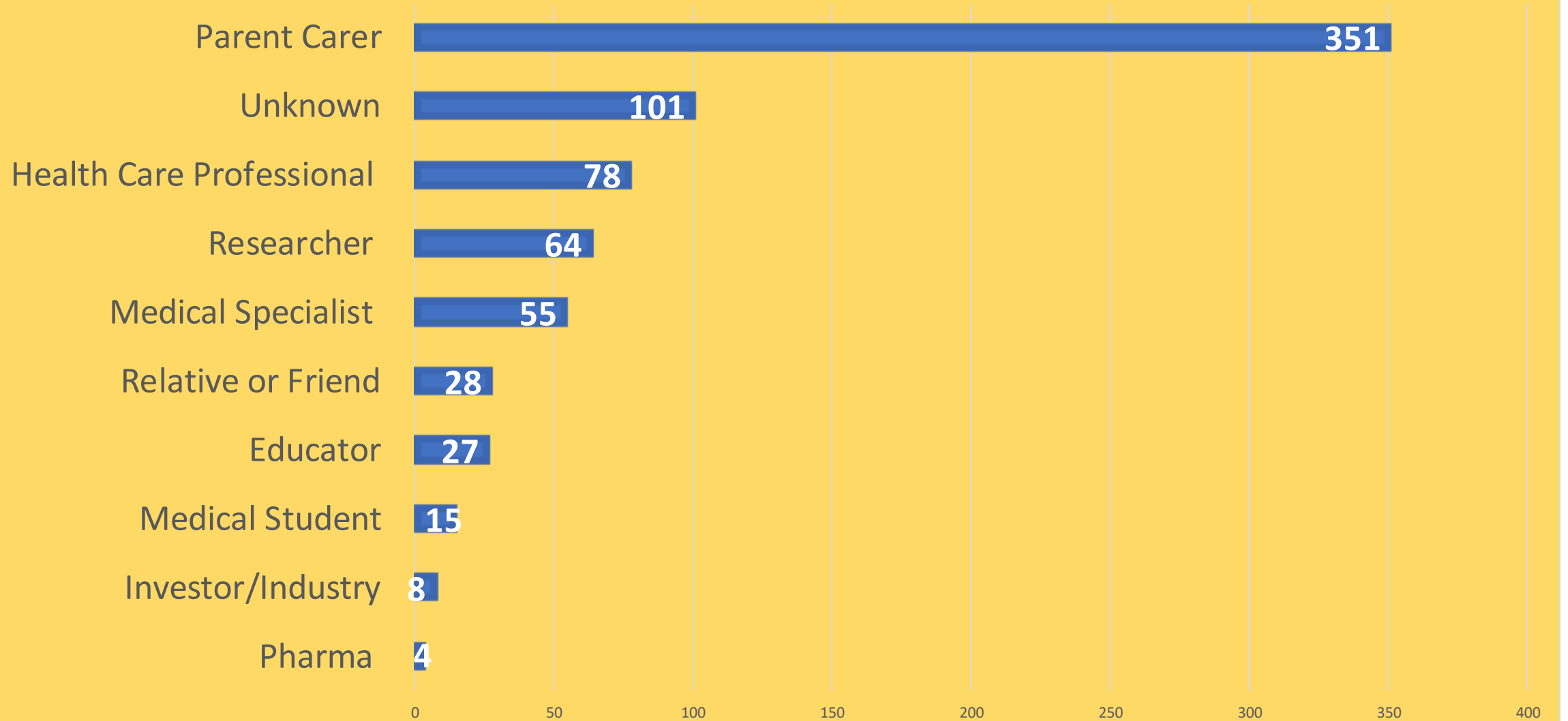
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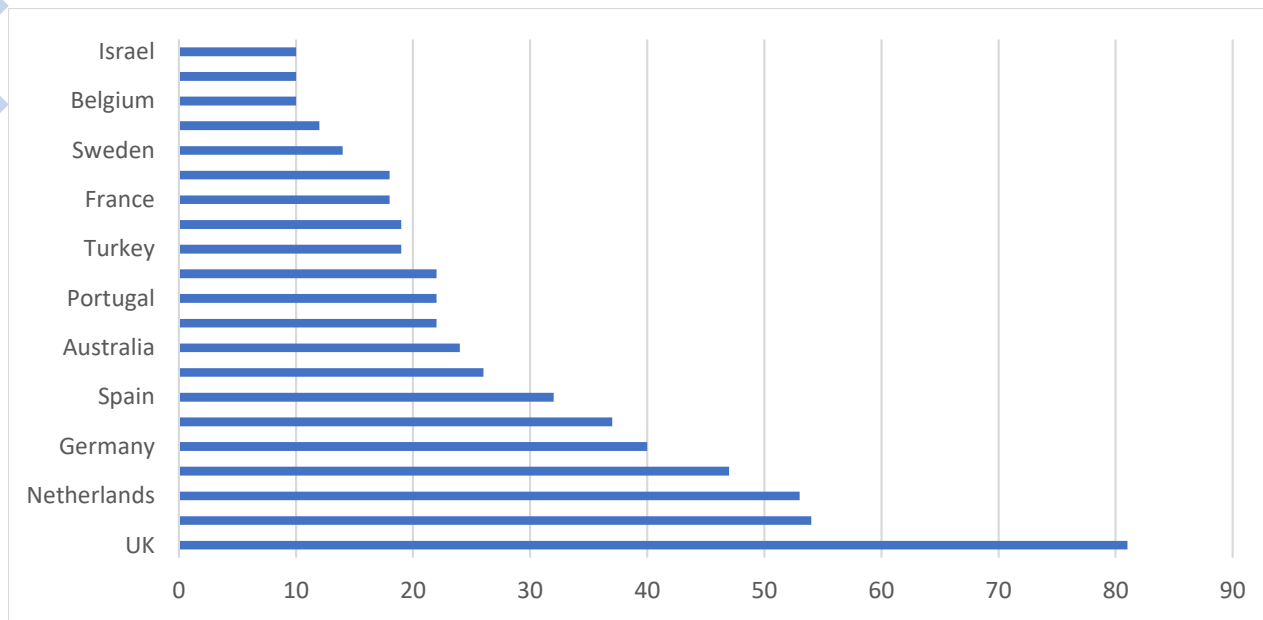


09:00 - 09:15	Re-cap and Introduction	Becky Jenner
09:15 - 09:30	I Can't Get No Sleep...	Karen Spruyt
09:30 - 09:50	Epilepsy in Rett Syndrome	Aglaia Vignoli
09:50 - 10:05	Control of Breathing	Ana Abdala
10:05 - 10:25	Tea Break	
10:25 - 10:40	Gastrointestinal Myths and Misconceptions in Rett Syndrome	Kathleen Motil
10:40 - 11:00	Identifying and Managing Emotional, Behavioural Autonomic Dysregulation in Rett Syndrome	Paramala Santosh
11:00 - 11:30	Discussion in Country Groups	
11:30 - 12:15	Q&A to Speaker Panel	
12:15 - 13:15	Lunch / Networking	
13:15 - 13:25	Physical Therapy Interventions in Rett Syndrome	Meir Lotan
13:25 - 13:35	Communication Research and the Rett Syndrome Communication Guidelines	Gillian Townend
13:35 - 13:45	Communication Continued: Well-being and Emotional Competence	Helena Wandin
13:45 - 14:00	Occupational Therapy: Role in Rett Syndrome	Pamela Diener
14:00 - 14:10	Music Therapy and People with Rett Syndrome	Linn Johnels
14:10 - 14:40	Discussion in Country Groups	
14:40 - 15:00	Tea Break	
15:00 - 15:30	Q&A to Speaker Panel	
15:30 - 16:00	Action Planning in Country Groups	
16:00 - 16:45	Feedback to Whole Group	
16:45 - 17:00	Summary, Thank You and Closing	Becky Jenner & Gillian Townend



VARIOUS ROLES





Number of participants by country (over 10)

Other countries represented were:

Denmark

Finland

Greece

Switzerland

Bulgaria

Ireland

Bosnia and Herzegovina

Botswana

New Zealand

Ukraine

Argentina

China

Cyprus

Egypt

Lithuanian

Montenegro

North Macedonia

Norway

Romania

South Africa

South Korea

Colombia

Ecuador

Estonia

Luxemburg

Macedonia

Malta

Russia

Singapore

Slovenija



Feedback and Outcomes

"Approaches to and advances in the research were most informative and really exciting. The topic was thoroughly covered and presentations of a very high calibre. For me, they reinforced the knowledge I had picked up over the decades and acted as a reminder of what can be achieved, and an encouragement to keep pushing the boundaries. I was heartened by all the positivity, the possibilities, and the evidence to support them.

"Pamela Diener's presentation was a real stand-out for me. I learned a lot from her methodical approach, and intend to put her recommendations into practice. Meir Lotan too had a fascinating presentation."

"The talks were very instructive and encouraging, and the speakers a joy to listen to. My sincere thanks go out to all of them, as well as the organisers, for their interest, professionalism and dedication to the task."

"This format of short online presentations combined with online and country Q & A is a great way to participate in this meeting. Compliments for the organization, especially for the subtitles. That makes it more easy for people to understand what is being said. Also, the fact that the presentations stay available on the website is great. In this way the caregivers for my daughter can see these presentations on their own time. We also use the presentations as a way of schooling them on the subject of Rett syndrome. "

Turkey

The event had **3 impacts** on the community that were very important:

- ★ First-hand access to more comprehensive information about the drug Trofenitide than they have ever had. Winnie and Prof. Dr. Yasemin Alanay from Acibadem University speeches helped the community reach a consensus on the need for them to be in close contact with the government on access to possible treatments, and to quickly establish an association so that they can be informed quickly of all developments, as was the case with this event.
- ★ They contacted the officials of the Rare Diseases Federation, which was established with the support of the Rare Diseases Center within the body of Acibadem University. With legal assistance and help of them, now we are preparing the establishment application of the Turkish Rett Syndrome Association.
- ★ They create several working group, one to establish the association, one to prepare webinars about AAC, PT and nutrition, and one to prepare documentation that will inform healthcare professionals about the commercial drug and especially the Taysha gene therapy trial.

They also prepared a comprehensive abstract report of the event which is now available on their website and shared this lovely quote with us from a Dad. <https://rettsendromu.com/index.php/etkinlik-sonuc-bildirgesi/>

After the Taysha presentation, one of the Rett dad said that “When my daughter was diagnosed in 2010, he asked "Is there any hope?". Doctor said, it took 20 years from Andreas Rett's early studies to name the disease, and an additional 20 years to find out which gene it was from... I came here without any hope at my wife's insistence, and now I can't stop myself from excitement because I learned that there is hope.

Georgia



Developing Rett Registry/Database including more information to be captured in it. Be able to find new/old cases of Rett, targeting neurological clinics/hospitals, daycare centers, etc.



Rett registry is already in place with the short information on the individuals with Rett.



Access to new drugs. As Georgia is not yet part of the EU/EEU what steps have to be taken to be able to have a registered drug for Georgian patients.



Trofinetide registration process is started between the Georgian association and ministry of health.



Genetic testing for all Rett patients and finding sources/funding to be able to cover the expenses for all undiagnosed patients.



They have had communication with the laboratory Blueprint to see what can be done with the patients who are in need of genetic testing. They had the program for developing countries but unfortunately, they terminated the project. So this remains an current action.

Serbia

After several long discussions, they agreed that their priorities are:

- ★ Creating adequate resident units for older girls
- ★ Achieving adequate medical care for girls, especially older ones
- ★ Creating new day care centres (existing centers are only in Belgrade, Novi Sad and Niš)
- ★ Engaging with new families, re-creating (after ten years) a network of parents and professionals
- ★ Planning an event/association meeting, with the attendees from this meeting and with families with Rett children, where they can discuss news and make plans for future.

Hungary

- ★ They have been invited from Bethesda children's hospital in Budapest to discuss a multidisciplinary Rett centre there for the pediatric population. They already had one meeting with the head of the neurology department where they expressed their needs. They will have the next meeting soon and they hope to have more concrete news in the next few weeks.
- ★ Sagi Judit who is working for the Rare disease institute in Budapest has been working for a while now in cooperation with Daniela Tropea on the preparations to start a Rett registry. At the moment she is working on the translation of the questionnaire and on getting licenses and approvals from the ethics and other committees in order to be able to start the project.
- ★ They revived their contacts with the diagnostic centre in Pecs and sent them a letter about the association that they promised to send to all the neurologists whose patients get Rett diagnosis. The idea is that they inform the parents about the parent association. The diagnostic process is different now. In most of the cases they seldom meet the newly diagnosed girls parents in person. Instead, they receive blood samples by delivery and in the case of the positive result inform the neurologist by post.
- ★ They are also in contact with Gill Townend and will be working on the translation of the Communication Guidelines.

United Kingdom

The UK already has a well-established parent association, Rett UK and a separate research charity, Reverse Rett with whom Rett UK have an alliance to collaborate on projects that benefit the whole community. Rett UK was one of the satellite meeting hosts and used this opportunity to invite students and trainees to learn more about Rett syndrome and hopefully develop new interests from future researchers and clinicians.

Rett UK post event aims to:

- ★ Continue to provide excellent support services in particular to Include access to information, resources, and support groups to help them cope with the challenges associated with the disease. The charity should also work to advocate for policies and programs that improve the quality of life for those affected by the disease, such as access to healthcare and social services. Consider platforms and resources that can easily be modified and changed
- ★ Continue to raising awareness amongst professionals particularly to identify people that are undiagnosed. Explaining what we do – diagnosis, benefit system etc... Increasing critical mass by joining with other rare disease charities where appropriate – digestive issues/ breathing problems... this is not just about Rett Syndrome. Look for existing organisations. Approach medical schools and the possibilities of including it in their curriculum – good start.
- ★ Working with Reverse Rett to prepare the UK for the future treatments both pharmacological and gene therapy.

Question: Who should be involved? All – Families. Other countries

And what are your next steps? Share plans and resources with the other organisations through increased networking and alliances.

Croatia

- More research is needed in Croatia but the meeting provided excellent opportunities to network with experienced researchers and clinicians. Professionals in Croatia are prepared to travel to gain experience and training in Rett syndrome.
- Croatian clinicians are really interested to learn more about a personalised, holistic approach to treating people with Rett syndrome.
- There is a recognised need to reach more families and to have a Rett registry.
- Improvements are needed in the infrastructure in Croatia to support better access to therapies in particular.
- Translation of the International Communication Guidelines in Rett syndrome is on their action plan too.



“To conclude this unique meeting has been very constructive and rewarding for Croatian Rett community but also for the entire country. All the stakeholders agreed that there should be more events like this in the future, and that this type of collaboration, understanding and mutual help that every attendee experienced during this wonderful event can indeed make a great difference in lives of Rett patients and their families in Croatia but also in all the other countries.”

Netherlands

The Netherlands have an established parent association; Dutch Rett Syndrome Association/Nederlandse Rett Syndroom Vereniging (NRSV) since 2008. NRSV focusses on providing support and knowledge for Rett families, support for research through international collaborations and works closely with the Maastricht Rett Expertise Centre. NRSV hosted one of the satellite meetings attended by parents and professionals.

NRSV post event aims to:

1. Continue to provide information and support, share and clarify international research results, explore the next steps in recent developments in Rett syndrome particularly in respect of treatment for adults, new drugs and on enhancing cognitive development. We will also continue to support research programs on movement and scoliosis and seek more international collaborations in this field.
2. Continue to work with international organisations and the Maastricht Rett Expertise Centre on future pharmacological treatments and gene therapy for Rett.
3. Share more knowledge and experience locally and with other countries e.g. with the networks of professionals in physiotherapy, communication and education. Share more plans and resources with other Rett associations internationally and with other rare disease organisations locally to create a stronger and more creative basis for everyday care and treatments. Expand collaborations for online information, both new and existing with subtitles in multiple languages.