

Rett Syndrome Europe

Activity Report 2018-2019

Tampere, Finland – 28/09/2019



RSE Board members



Danijela Szili Yvonne Milne Wilfried Asthalter Stella Peckary Caroline Lietaer

Other volunteers: our former presidents Gerard Nguyen, Thomas Bertrand
Newly introduced volunteers: Tim Addison, Laura Kanapieniene, Tim Lang

RSE aims of the statutes

- **AIM 1:** To make Rett syndrome better known in all European countries to the public, professionals, carers and those who are directly concerned
- **AIM 2:** To improve the communication within the European Rett Community
- **AIM 3:** To promote as a representative European organisation, the interests of people with RTT and families
- **AIM 4:** To expand RSE to all European countries and to assist, if necessary, in the creation of national associations
- **AIM 5:** To promote research into Rett syndrome

Building the Network

To make Rett syndrome better known to the public, professionals, carers

News/Articles/Events on RSE website

Report of international congresses

Family days in the different countries, seminars

Calendar of webinars organised by US rettsyndrome.org

Rett Resource project



An invaluable resource, written for you by European Rett experts,
with a focus on therapies and advice on day to day care and management of the complex problems of Rett syndrome.

- An invaluable resource and substantial guide with knowledge for families, therapists and other people who are involved and interested in supporting and assisting people with Rett syndrome
- Choice of the authors made by Rett Expertise Centres and European Rett Associations

Rett Resource project

Rett Resource in progress



- Romanian
- Bulgarian
- Arab?
- Persian?

German

French

- English
- Lithuanian
- Croatian
- Dutch
- Macedonian(3)
- Serbian
- French
- German
- Russian
- Turkish

Rett Disorders Alliance Health Checklist

New resource to support the best treatment and care for people with Rett syndrome and FOXP1

Rett Disorder Alliance is an alliance between Rett UK, Reverse Rett and FOXP1

Download on: <https://www.rettuk.org/resources/resources-for-families/rett-disorders-alliance-health-checklist/>



Improve the communication

– With the Rett Associations:

- Constant update of all contacts
- Website: www.rettsyndrome.eu
- Facebook page and closed group
(Closed group makes it possible to contact the representatives and some experts)
- Rett community on rareconnect platform: www.rareconnect.org

– With the Rett Expertise centres

– Helping the Rett Associations outside Europe

- Newly created associations in Tunisia and Morocco (can be observers)

Building the Network

The newly created Rett Centre in Belgium

- Multidisciplinary Consultation and follow-up for children and adults with Rett syndrome and MECP2 duplication syndrome- UZ Leuven

- Prof. Hilde Van Esch, clinical geneticist
- She described the MECP2 duplication



- Dr. Katleen Ballon, rehabilitation paediatrician-rehabilitation

Building the Network

Dr Gvantsa Kvantaliani from Georgia received as doctor and Rett mother a training in the [Rett Expertise Centre of Maastricht](#). She is starting up an association in her country. We express our gratitude to prof. Leopold Curfs for making her stay possible



Building the Network

Caroline represented RSE on the PHD defence of **Gillian Suzanne Townend**

“Rett syndrome: Recognising the Communication Challenges, Needs and Potential of Individuals Living with a Rare Disease”

Thanks to Netherlands Rett Expertise Centre, NRSV, Stichting Terre and Rettsyndrome.org. RSE helped by translating and diffusing the questionnaires



Building the Network

7th South East European Rare Disease Conference 7-9 June, 2019 in Struga, North Macedonia

- At the same time two other meetings took place: scientific meeting and Gaucher regional meeting. It was organized by [Life With Challenges / Живот со Предизвици](#)
- Around 300 participants mostly from the Balkan countries took place.
- Danijela Szili represented Rett Syndrome Europe with the talk about what is there, when treatment is not existent and held a workshop for Rett families from Macedonia.



External Influence and Advocacy

To promote as a representative European organisation, the interests of people with RTT and families

→ *Having RSE officially in the network of European institutions*

1. **RSE is a member of EURORDIS:** Allows RSE to vote at the GA of EURORDIS During the ECRD (European Congress for Rare Diseases), we can apply for Eurordis Fellowships to support our members to attend our meetings and Eurordis/ECRD annual meeting
2. **RSE is involved in ITHACA/ERN :** RSE board member Yvonne is a rare disease European patient advocate in the European Reference network ITHACA. She represents Rett syndrome and other rare diseases in this important forum. Following Brexit , ITHACA now co-ordinated in France by Alain Verloes and his team



External Influence and Advocacy

3. EURORDIS Winter school (Danijela Szili): March 2019

- The training was about scientific innovations and translational research and was held in the prestigious Imagine Institute in Paris. Inspirational lectures, interactive sessions between participants and visits to the laboratory and animal facility of the Imagine Institute.



External Influence and Advocacy

4. EURORDIS Task Force (Danijela Szili):

- DITA Drug Information and Transparency Access Task Force

5. EMA European Medicines Agency (Danijela Szili)

External Influence and Advocacy

7. Rett Networked Database (Yvonne Milne and Danijela Szili):

- Webinar organised on 8/02/2019
 - Attended by 8 countries
- Yvonne and Danijela are parent representatives on the Scientific Committee
- Attend regular Skype meetings with the Network
- Give parent perspective for EU grant applications eg 2019 European Joint Programme on Rare Diseases
- Encourage and Facilitate collaboration between Networks using databases, eg Australian InterRett

Our Network

To expand RSE to all European Countries and to assist, if necessary, in the creation of national associations

Ukraine : Ukrainian Rett Syndrome Association was founded 08.05.2018 in Kiev. There are 40 members in the association and the community.

Turkey: The Rett syndrome community in Turkey comprises 30 families, under the leadership of 9 families who have revived the Turkish Rett Syndrome Association, which was originally established in 2002, but ceased activities in 2015. The number of families that the community is connected to is over 100. The community publishes an e-magazine called ogRETTim (meaning: education) www.rettsendromu.com

Georgia: Recently established Georgian Rett Syndrome and other Rare Diseases Association was founded in June 2019 by 2 Rett families. This Association includes Rett families as well as medical professionals. In Georgia, we assume to have around 50 Rett families. Our mission in following period are to make Rett syndrome better known in Georgia to the public, professionals and carers, bringing families together and share knowledge.

Our Network

To expand RSE to all European Countries and to assist, if necessary, in the creation of national associations

– Outside Europe (accepted as observers)

Morocco: the association has been created on 19/12/2015 by Mr El Mokhtar Mustapha in Temona. The association has 60 people with RS. For more info: ams.rett@gmail.com

Tunisia: the association of Angelman and Rett syndrome(ATSAR) has been created in 2018 by doctors and parents. For the moment 46 people with RS are known. For more info: <https://atsar-tn.org/>

Research

To promote research

We post/publish/diffuse the call for grants of our members (Finrett)

This year our help was also requested for:

- **Study to increase physical activity in Rett Syndrome**
Michelle Stahlhut ,Denmark/Cochavit Elefant and Meir Lotan, Israel
Helen Leonard,Australia
- **In the European Joint programme on Rare Diseases Consortium EJP RD JTC2019 with:**
Sleepstudy of Rett and Angelman syndrome Karen Spruyt
Pa CIP-ID Networked Database Alessandra Renieiri

RSE research subcommittee

Coordinator from the RSE board: Szili Danijela

10 volunteers from 8 countries

Scope:

- ❖ Mapping labs, hospitals and researchers involved in RTT research, starting with the research conducted in Europe.
- ❖ Both medical and genetic research as well as clinical trials will be included.
- ❖ Only genetic research involving MECP2 gene will be taken into account.

RSE research subcommittee

Tasks

- ❖ Searching published articles from 2014 in medical and scientific journals available online
- ❖ Creating a list of RTT research categorized by: (country, type of research (genetic, medical, interventional or non-interventional), location and the date as well as the name of the scientific or medical journal where it was published.
- ❖ Creating a list of scientists and physicians working on RTT categorized by country, type of research and location (name of the hospital, university, public or private institute).

RSE research subcommittee

Country	Type	Type II	Subject	Subject II	Location	Published at	Year	Publication link	Author
2 Austria	Clinical		Response to name		Medical University Graz	Research in Developmental Disabilities	2018	https://doi.org/10.1016/j.ridd.2018.04.01	Peter B Marschik
3 Austria	Review		Biomarkers	Regression biomarkers	Medical University Graz	Neuroscience & Biobehavioral Reviews	2019	https://doi.org/10.1016/j.neubiorev.2019.04.001	Peter B Marschik
4 Austria	Clinical		Vocalisations		Medical University Graz	Research in Developmental Disabilities	2018	https://doi.org/10.1016/j.ridd.2018.02.001	Peter B Marschik
5 Austria	Clinical		Biomarkers	Early behavioural biomarkers	Medical University Graz	Wiener Medizinische Wochenschrift	2016	https://doi.org/10.1007/s10354-016-049-0	Peter B Marschik
6 Austria	Clinical		Behavioral ?????		Medical University Graz	Neuroscience & Biobehavioral Reviews	2019	https://doi.org/10.1016/j.neubiorev.2019.04.001	Peter B Marschik
7 Austria	Clinical		Preserved Speech Variant		Medical University Graz	Developmental Neurorehabilitation	2014	https://doi.org/10.3109/17518423.2013.811111	Peter B Marschik
8 Austria	Clinical		Early symptoms	Early symptoms in twins	Medical University Graz	Journal of Developmental and Physical Disabilities	2014	https://doi.org/10.1007/s10882-013-935-0	Peter B Marschik
9 Austria	Review		Vocalisations		Medical University Graz	Advances in Neurodevelopmental Disabilities	2018	https://doi.org/10.1007/s41252-017-005-0	Peter B Marschik
10 Austria	Clinical		Gall bladder		University of Vienna	Dev Med Child Neurol	2014	https://doi.org/10.1111/dmcn.12358	Michael Freilinger
11 Norway	Clinical	Single case re	Epilepsy	Genetics/SCN1A gene	Drammen hospital	BMC Medical Genetics	2018	https://doi.org/10.1186/s12881-018-070-0	Mari Wold Henriksen
12 Norway	Clinical		Epilepsy	Genetics	Drammen hospital	Epilepsy Research	2018	https://doi.org/10.1016/j.eplepsyres.2018.04.001	Mari Wold Henriksen
13 Sweden	Clinical		Daily activities		National Center for Rett syndrome	Scandinavian Journal of Occupational Therapy	2018	https://doi.org/10.1080/11038128.2018.1481111	Asa-Sara Semhelf
14 Sweden	Clinical		Daily activities		Swedish National Rett Center, Öste	Scandinavian Journal of Occupational Therapy	2016	https://doi.org/10.1080/11038128.2016.1181111	Asa-Sara Semhelf
15 Sweden	Clinical	Single case re	Physiotherapy		National Center for Rett syndrome	Wiley Online Library	2013	https://doi.org/10.1002/ccr3.2013	Lena Svedberg
16 Sweden	Clinical		Physiotherapy	Treadmill	National Center for Rett syndrome	Research in Developmental Disabilities	2018	https://doi.org/10.1016/j.ridd.2018.08.001	Gunilla Larsson
17 Sweden	Clinical		AAC communication		Uppsala University	IOS Press Ebooks			Helena Wandin
18 Sweden	Clinical	Focus group	AAC communication		Uppsala University	Disability and Rehabilitation	2014	https://doi.org/10.3109/09638288.2014.911111	Helena Wandin
19 Denmark	Clinical	Focus group	Functional mobility scale		Center for Rett Syndrome, Glostrup	Disability and Rehabilitation	2017	https://doi.org/10.1177/2F104973231881111	Michelle Stahlhut
20 Denmark	Clinical		Physical activity		Center for Rett Syndrome, Glostrup	Disability and Rehabilitation	2017	https://doi.org/10.1080/09638288.2017.1311111	Michelle Stahlhut
21 Denmark	Clinical		Functional mobility scale		Center for Rett Syndrome, Glostrup	Disability and Rehabilitation	2016	https://doi.org/10.1080/09638288.2016.1211111	Michelle Stahlhut
22 Denmark	Basic		Male Mosaicism		Center for Rett Syndrome, Rigshosp	Clinical genetics		https://doi.org/10.1111/cge.13473	Bitten Schönewolf-Gr
23 Denmark	Review		Danish cohort		Center for Rett Syndrome, Rigshosp	Clinical genetics		https://doi.org/10.1111/cge.13153	Bitten Schönewolf-Gr
24 Denmark	Clinical		Aging	Functional ability	Center for Rett Syndrome, Rigshosp	Disability and Rehabilitation	2016	https://doi.org/10.3109/09638288.2016.1111111	Bitten Schönewolf-Gr
25 Denmark	Review		Diagnostics	Danish diagnostics	Center for Rett Syndrome, Glostrup				Bitten Schönewolf-Gr
26 Slovenia	Clinical		Transcranial magnetic stimulation		University Medical Centre Ljubljana	European Journal of Pediatric Neurology	2016	https://doi.org/10.1016/j.ejpn.2016.03.001	Natalija Krajc
27 Slovenia	Review		Epilepsy	Treatment	University Medical Centre Ljubljana	Dovepress		https://doi.org/10.2147/TCRM.S55896	Natalija Krajc
28 Slovenia	Clinical	Single case re	Respiratory dysrhythmia	Topiramate	University children's Hospital Ljubl	Journal of Child Neurology	2015	https://doi.org/10.1177/0883073813508111	Natalija Krajc
29 Portugal	Basic	Animal/mous	Bone health	Biomechanical properties	Faculdade de Engenharia da Univ	Bone	2015	https://www.sciencedirect.com/science/article/pii/S0021929015001111	Diogo Constante
30 Portugal	Basic	Cell	iHSPCs	Neural commitment	Department of Bioengineering and	Biotechnology Journal	2015	https://online.library.wiley.com/doi/10.1002/biot.201500111	Tiago G. Fernandes
31 Portugal	Clinical	Single case re	Epigenetic changes and over	Pericentric X-chromosome	Neurology Department, Hospital Dc	International Journal of Developmental Disabilities	2015	https://www.sciencedirect.com/science/article/pii/S0021929015001111	José Pedro Vieira
32 Belgium	Basic	Cell	Xi reactivation	Mixed modality approach	Center for Medical Genetics, Ghent	PNAS	2017	https://www.pnas.org/content/115/4/E1111111	Lieslot L. G. Carrette

Rett Centres Survey

- **AIM:** To collect data about different types of Rett Centres all across Europe, summarize the information and present it to Rett families and specialists.
- **Targeted Rett Centres:** Medical, Therapy and Research centres that have RS patients as their main target group or have *sufficient expertise* in the field.
- **Form of the survey:** [An online questionnaire](#) sent out by email to targeted Rett Centres and filled in by their representatives (only in English).
- **Results:** Submitted data will be organized and published on RSE website, insights discussed during the next GA.
- **Coordinator:** Laura Kanapieniene laura.kan@outlook.com

Rett Centres Survey

- The questionnaire has been sent out to our Member Associations to distribute it further for the Rett Centres, but the response rate was low. (*I wonder if my emails went to spam??*)
- Some Rett Centres were contacted directly
- We have responses from:
 - Swedish National Center for Rett syndrome and related diseases
 - Child Neurology, Medical University Hospital Vienna
 - Evelina London Children's Hospital
 - Centre de référence Marseille