

Rett Syndrome Europe

Activity Report 2017-2018

Copenhagen – 26/10/2018



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

RSE statutes changes

- **TWO MAJOR CHANGES**

- 1/ Art 10 : At least four members (instead of two) must be family or carers of a person with Rett Syndrome
- 2/ Term of an elected board member will be three years instead of two. They may be elected for a further two terms.

- **MINOR CHANGES:**

- 1/ Name and addresses of board members
- 2/ Address of registered office: financial consequences
- 3/ Article numbers • Dates of amendment laws

Building the Network

- **AIM 1:** To make Rett syndrome better known to the public, professionals, carers [...]

Inform, advertise special events

News/Articles/Events on RSE website

Report of international congresses

Family days in the different countries, seminars on certain Rett topics

Newly added is the 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
- Available in English, Croatian, Dutch and Lithuanian. Coming soon in French, German

Rett Resource project

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[Drooling management](#) – Helen Cockerill, UK

[Emotion, behaviour and depression](#) – Sarojini Budden, USA

[Epilepsy in Rett syndrome](#) – Nadia Bahi Buisson, Marie Hully, Elisabeth Celestin, FR

[Genetic aspects of Rett syndrome](#) – Laurent Villard, FR

[Hippotherapy: developing motor and communication skills](#) – Lena Svedberg, SE

[Hydrotherapy](#) – Stella Peckary, AT

[Management of gastrointestinal disorders in Rett syndrome](#) – Jenny Downs & Helen Leonard, AU

[Managing bone health in Rett syndrome](#) – Jenny Downs & Helen Leonard, AU

[Managing poor growth and nutrition in Rett syndrome](#) – Jenny Downs & Helen Leonard, AU

[Managing scoliosis in Rett syndrome](#) – Jenny Downs & Helen Leonard, AU

[Music therapy: meeting physical, emotional, cognitive and social needs](#) – Märith Bergström-Isacsson, SE

[Nutrition and hydration](#) – Irène Benigni, FR

[Occupational therapy for hands: facilitating adults' daily living activities](#) – Asa-Sara Sernheim, SE

[Physical therapy for Rett syndrome](#) – Meir Lotan, IL

[Physiotherapy: developing and improving motor functions](#) – Lena Svedberg, SE

[Puberty in Rett syndrome](#) – Hilary Cass UK

[Scoliosis in Rett syndrome](#) – Sabine Leimer AT

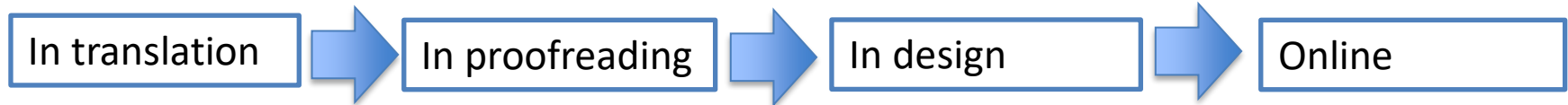
[Screaming episodes in Rett syndrome](#) – Hilary Cass, UK

[Sleeping problems](#) – Lietaer Caroline & Robert James Goddard, NL

[Stereotypies and movement disorders in Rett syndrome](#) – Nadia Bahi Buisson, Marie Hully, Elisabeth Celestin, FR

Rett Resource project

Rett Resource in progress



- French
- Russian

- Croatian
- Dutch
- Macedonian

- Dutch

- English
- Lithuanian
- Croatian (5)
- Dutch (2)
-

Rett Resource project

Our aim for the future is to have Rett Resource translated with your help into all European languages and provide them on our website www.rettsyndrome.eu.

A fundraising flyer has been created by Andreas Meissner, Laura Kanapieniene and Laura Luisaityte. This can help you to have Rett Resource translated into your country`s language. They created this banner with the aim of enabling other national associations to adapt and use it to fundraise for the project in their own home country



The flyer features the RSE logo (two hands holding stars) and the text "rse rett syndrome europe". A large yellow banner reads "RETT RESOURCE Support translation into Lithuanian language". To the right is a 3D rendering of a box labeled "RETT RESOURCE" with a colorful dot pattern. At the bottom left is the Agotos logo with the text "Agotos Inžinierius ir paramos fondas". At the bottom right, donation details are provided: "Donate to: AGOTOS LABDAROS IR PARAMOS FONDAS code: 304090279 iban: LT507044060008049810 swift: CBVILT2X".



Rett Resource project

General terms and conditions to translate the Rett Resource.

- We have guarantee to the authors that translations can only be done by professionals.
- After translation it has to be proofreaded by someone which knows very well Rett syndrome
- This is the most important phase
- After the proofreading it has to be send to caro.lietaer@skynet.be so we can put the articles in our design to publish it on the website.
- You can't publish it on your own website (it's in the contract with the authors) but you can put a link from your website towards our website.
- You will find in annexe the general file with all articles and the number of words which can help you to obtain prices in translation offices.
- References need no translation, so do not waste time and money on this , do not forget the translation offices to tell there is no need to translate them.

Building the Network

- **AIM 2:** To improve the communication within the European Rett Community
 - With the Rett Associations: constant update of all contacts in all the countries
 - Website: www.rettsyndrome.eu
 - Facebook group (open and closed group): www.facebook.com/Rett-Syndrome-Europe
 - Rareconnect platform: www.rareconnect.org
 - Newly created Associations : Turkey, Tunisia, Ukraine
 - With the Rett Expertise centres
 - Enabling meeting between the Rett Centre Paris and Rett Centre of Maastricht

Building the Network

- The newly created Rett Centre in Paris



- Created in 2017 in Paris, the reference centre is coordinated by **Prof. Nadia BAHIBUISSON** and welcomes people with multiple disabilities related to developmental encephalopathies such as Rett syndrome, related genetic syndromes (CDKL5, FOXP1), syndromes of Aicardi, Angelman and brain malformations.

French protocole

<http://hopital-necker.aphp.fr/wp-content/blogs.dir/14/files/2017/04/PNDS-Rett.pdf>



Protocole national de diagnostic et de soins (PNDS)

SYNDROMES DE RETT ET APPARENTÉS

Texte du PNDS

Centres de référence maladies rares

Déficiences intellectuelles de causes rares
Filière DéfiScience

&

Anomalies du développement et syndromes
malformatifs de l'interrégion Est
Filière AnDDI-Rares

Avril 2017

Centre de référence Déficiences intellectuelles de causes rares
Centre de référence des Anomalies du développement & syndromes malformatifs de l'interrégion Est
Avril 2017

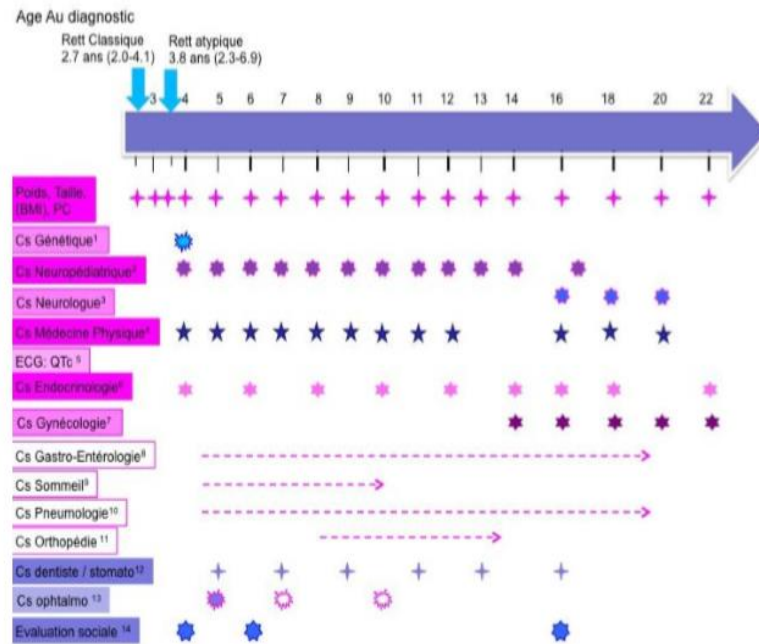


Figure : La grande flèche bleue indique l'âge de l'enfant (en années) au cours du suivi. L'âge médian au diagnostic du syndrome de Rett classique : 2,7 ans (intervalle 2,0-4,1) et l'âge médian au diagnostic du syndrome de Rett atypique 3,8 ans (intervalle 2,3-6,9) sont indiqués par une flèche verticale bleue.

Le suivi de la croissance (poids, taille, BMI et PC) se fait tous les 6 mois jusqu'à 4 ans puis tous les ans jusqu'à 14 ans, puis tous les 2 ans ; l'objectif de ce suivi est d'identifier une cassure de la courbe pondérale et/ou staturale. L'objectif de BMI est au 25^e percentile.

Building the Network

Collaboration meeting between Rett Centre Paris en Maastricht Expertise Centre Maastricht (Lietaer Caroline)

Representatives of the recently opened Rett Centre in Paris and the Maastricht Rett Expertise Centre meet together on 15/02/2018 in Paris to discuss new ways of working together on aging in Rett syndrome



Building the Network

- **The Catalan Rett Association celebrated their annual Congress in Barcelona "Rett: Abriendo puertas" the 7th of April (Caroline Lietaer)**
- This year it was focused on physical and cognitive learning capacities.
- There were present 120 people, most of them relatives, but also assistant doctors, teachers, physiotherapists, speech and music therapists and 320 people attended through streaming.
- There were presentations of Lotan Meir, Gill Townend and Caroline Lietaer



External Influence and Advocacy

- **AIM 3:** To promote as a representative European organisation, the interests of people with RTT and families
 - *Having RSE officially in the network of European institutions*
 - *Raising Awareness*
- 1. **RSE is a member of EURORDIS:** Allows RSE to vote at the GA of EURORDIS During the ECRD (European Congress for Rare Diseases),
- 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.

External Influence and Advocacy

3. EURORDIS Training resources (Yvonne Milne):

- Winter School – New initiative for training rare disease advocates - IMAGINE Institute, Paris, March 2018. A “Capacity building programme for **patient experts** on scientific innovation and translational research”



External Influence and Advocacy

4. EURORDIS Task Force (Danijela Szili):

- DITA Drug Information and Transparency Access Task Force

5. EMA European Medicine Agency (Danijela Szili):

- Scientific Advisory Groups meeting

SAGs are created by the CHMP (Committee for Medicinal Products for Human Use) to deliver answers, on a consultative basis, to specific questions addressed to them. The Committee, while taking into account the position expressed by the SAG, remains responsible for its final opinion.

External Influence and Advocacy

6. EFNA European Federation of Neurological Associations (Yvonne Milne):

- Yvonne gave a presentation about our work in Rett Syndrome Europe at EFNA's AGM in Lisbon in June. This meeting ran parallel with the Fourth EAN (European Academy of Neurology) meeting, attracting 6,700 neurologists from around the world.
- The EFNA meeting brought together European umbrella organisations of neurological patient advocacy groups, including the EAN, with a theme of "Partnership in Progress"

External Influence and Advocacy

7. ECRD Meeting Vienna (Yvonne Milne, Stella Peckary and Danijela Szili):

Organised by EURORDIS and co-organised by Orphanet and the DIA

European Conference on Rare Diseases & Orphan Products on 12/05/2018 in Vienna was an opportunity to discuss and reach solutions on how we can **look to the future** to improve the lives of the estimated 30 million people living with a rare disease in Europe and 300 million worldwide.



Our Network

- **AIM 4:** 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). For more info: www.rettsendromu.com

Tunesia: has a new association of Angelman and Rettsyndrome(ATSAR). ATSAR is invited as observer as they are the only and first association in the Arab countries. For the moment 46 people with RS are known. For more info: <https://atsar-tn.org/>

Research

- **AIM 5:** To promote research (Danijela)

ROME, SEPTEMBER 27TH-29TH, 2018

www.rettrome2018.org

Recent advances in basic, translational and clinical research on Rett syndrome. The 29th will be dedicated to parents and families and will include lay summaries of the scientific meeting. The report is available on our website



Statute changes and legal status of RSE

- ❖ **AIM:** Necessary statute changes that include new address, board members report and translation to French as requested by law in Lux.
- ❖ **Results:** We are a legal ASBL (association sans but lucratif) listed on the Luxembourg Business Register www.lbr.lu with the identity number of 20016102475 since 28 February 2018
- ❖ **Financial consequences :** Lawyer's expenses : For 2017: 877.50 euro, 2018: 595.78 euro and yearly domiciliation fee plus mail until now: 146.25 euro but will be 250 euro per year
- ❖ **Administrative consequences:** The management and administration of RSE as a European Federation and official ASBL in Lux has become much more complicated than in former years.

Proposal for RSE Subcommittees

❖ **AIM:** To find volunteers for Research and Awareness subcommittees. Their work would involve skype meetings once a month, e-mail communication and meetings during conferences. **Tasks** would be:

❖ **Research:**

- ❖ Research projects evaluation
- ❖ Rett Database Network
- ❖ Horizon scanning for new therapies
- ❖ Mapping labs and researchers involved in RTT
- ❖ Natural History Studies in Europe
- ❖ Help researchers communicate more
- ❖ Keep contact and cooperate with RTT research foundations
- ❖ Start involving scientists in the future as advisors and work on the creation of the EU Scientific board
- ❖ Help organization of scientific meetings and conferences
- ❖ Follow research news and make short reports for our Website and Facebook
- ❖ Create a Rett syndrome **CAB (Community Advisory Board)** when possible and attend EURORDIS trainings like Summer and Winter School; also use EURORDIS online learning tools for education

❖ **Awareness:**

- ❖ RSE Website, collecting and publishing articles and help with technical issues
- ❖ Post more on Facebook and be more active in October month, on Rare Disease Day and when there is a need
- ❖ Rare Connect,
- ❖ Twitter
- ❖ Creating banners

❖ **Coordinator:** Szili Danijela For all the questions and if interested to join write to szilidanijela@gmail.com

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