

Rett Syndrome Europe

Activity Report
2017-2018



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

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 (rettsyndrome.eu) at the top left. The main title is "RETT RESOURCE" in large blue letters, with the subtitle "Support translation into Lithuanian language" below it. A 3D rendering of the "RETT RESOURCE" book is shown on the right. At the bottom left is the logo for "Agotos" (Inėtaros ir paramos fondas). At the bottom right, the donation details are provided: "Donate to: AGOTOS LABDAROS IR PARAMOS FONDAS", "code: 304090279", "iban: LT507044060008049810", and "swift: CBVILT2X".

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

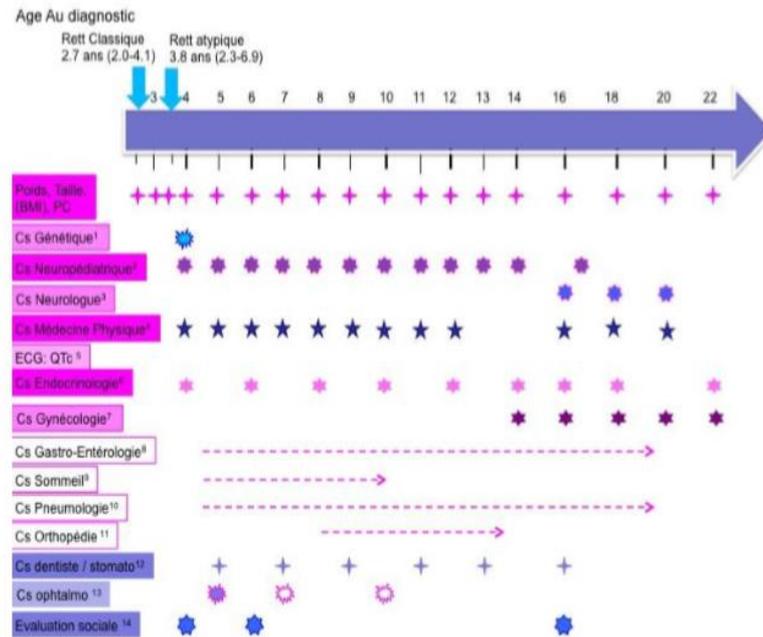


Figure : La grande flèche bleue indique l'âge de l'enfant (en années) au cours du suivi. L'âge médian au diagnostic du syndrome de Rett classique : 2,7 ans (intervalle 2,0-4,1) et l'âge médian au diagnostic du syndrome de Rett atypique 3,8 ans (intervalle 2,3-6,9) sont indiqués par une flèche verticale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

Rett patients in Europe



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 future

- Creation of subcommittees ?
- Questionnaire Rett Centres / Therapy Centres ?