Rett Syndrome Europe Activity Report

2015 - 2016

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RSE Board members (2016)

Caroline Lietaer

Yvonne Milne

Danijela Szili

Wilfried Asthalter

Thomas Bertrand

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RSE aims of the statutes

- **AIM 1:** To make Rett syndrome better known to the public, professionals, carers and those who are directly concerned in all European countries
- **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

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Aim 1: To make Rett syndrome better known to the public, professionals, carers […]

Thomas Bertrand at the 6th Catalan Rett day "The Future Begins Today" (Barcelona – May 2016)

During that day, patient representatives, researchers, medical doctors, directors of medical centres and officials from the Ministry of Health gathered in front of around 80 parents on site and hundreds connected via a live stream to connect families in Spain and other Spanish-speaking families around the world.

One part of the conference was focused on the place of Rett syndrome in the landscape of European Reference Networks.
The second part of the day focused on the situation of Research on Rett syndrome.

Aim 1: To make Rett syndrome better known to the public, professionals, carers […]

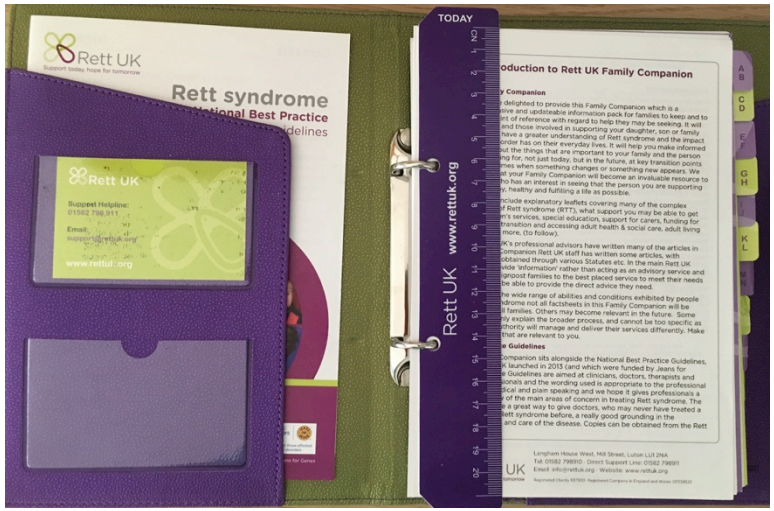
Caroline Lietaer: "Rett Resource"

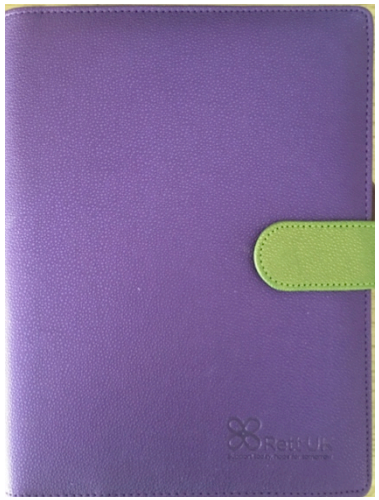
RSE has initiated a project of gathering information on a large panel of different topics under the form of RettUK's Family Companion. This project is not an update of UK's Family Companion but rather a reflection on international knowledge.

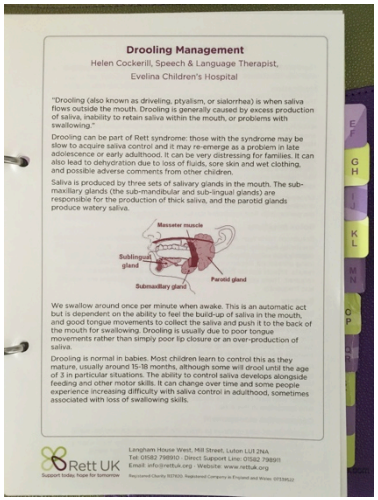
It aims at families and doctors to have a minimum information, hints about the disease and what to do depending on the situation.

This is not like medical guidelines as it should stay simple and as light as possible.

It is not written by RSE but by professionals around Europe carefully chosen by RSE representatives.







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Aim 1: To make Rett syndrome better known to the public, professionals, carers […]

Caroline Lietaer, Becky Jenner (RettUK) – New RSE Flyer

The aim of this project is to produce an eye-catching professional flyer about RSE and our activities.

In order to improve our visibility/ influence and to prepare for possible future fundraising.

The format(s) is not yet decided.

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AIM 2: To improve the communication within the European Rett Community

Since November 2015:

- Emails

  > 250 (e.g. Thomas)

- Rettsyndrome.eu

  14 articles, >1000 visitors

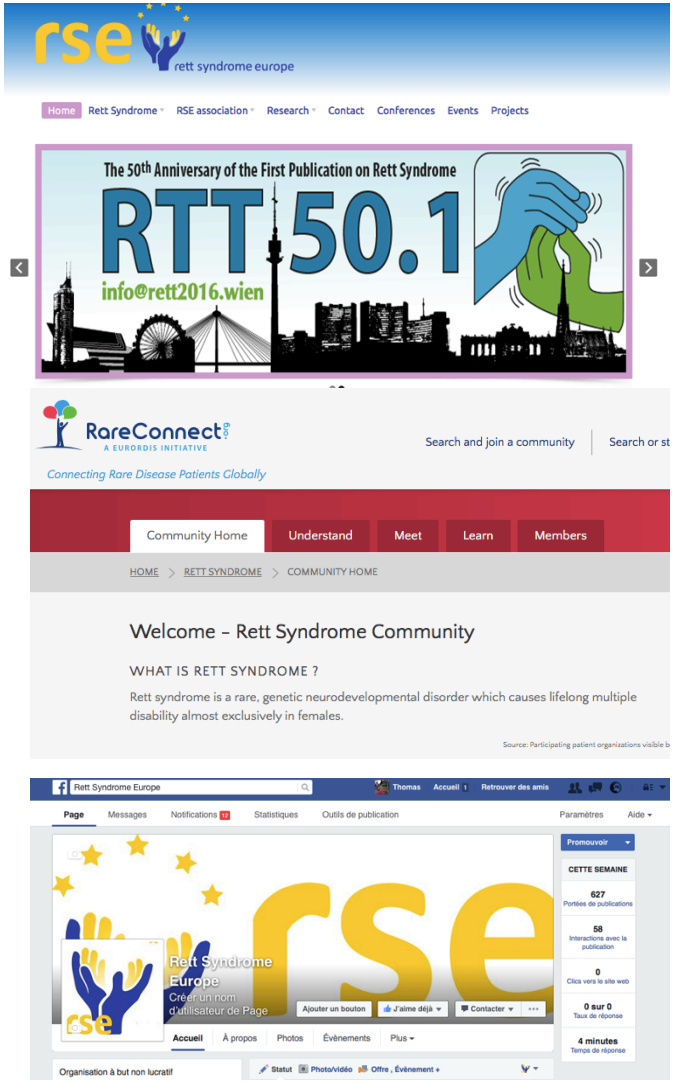
- RareConnect.org

  13 discussions, 336 members, >1000 visitors

  8 languages (Russian, Serbo-Croatian)

- Facebook

  public and private groups ~300 average views per post – peak July 16 of >1,5K views



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AIM 2: To improve the communication within the European Rett Community

- Actively contribute to international events on Rett syndrome
  - Discuss the choice of topics (parents, experts)
  - Discuss the choice of speakers
  - Financially (e.g. invited speakers, luncheons, invited representatives)

Rome

Kazan

Vienna

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AIM 3: To promote as a representative European organisation, the interests of people with RTT and families

- External Influence and Advocacy

  → *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 and Orphan products), *Danijela in Edinburgh (May 2016)*

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External Influence and Advocacy

2. EURORDIS Training resources:

- Summer School "A capacity building programme for patient representatives and researchers on information and access to orphan, paediatric, advanced therapies and health technology assessment."

  Laura Kanapieniene (Lithuania), Pedro Rocha (Catalonia) - June 2016, Barcelona *APPLICATION FOR 2017 SUMMER SCHOOL NOW*

3. EURORDIS Task Force:

- DITA Drug Information and Transparency Access Task Force (*Danijela Szili)*
  - *F2F Meeting in November 2015, Danijela re-elected for 3 more years. Friðrik Friðriksson (Iceland) was elected DITA member.*

4. EMA European Medicines Agency (London):

- Scientific Advisory Groups meeting – July 2016 (*Danijela Szili*)

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.

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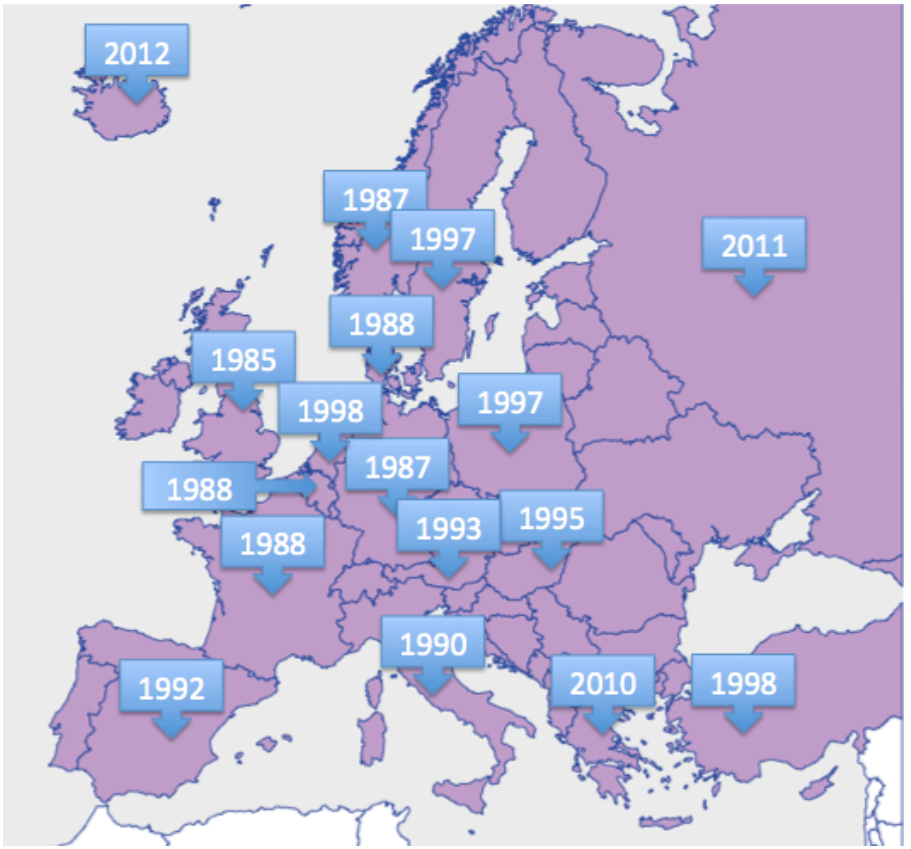
AIM 4: To Expand RSE to all European Countries and to assist, if necessary, in the creation of national associations

- ✓ Lithuania
- ✓ Belarus
- ✓ Kazakhstan
- ✧ Albania?
- ✧ Azerbaijan?
- ✧ Georgia?



→42 family associations or family contacts

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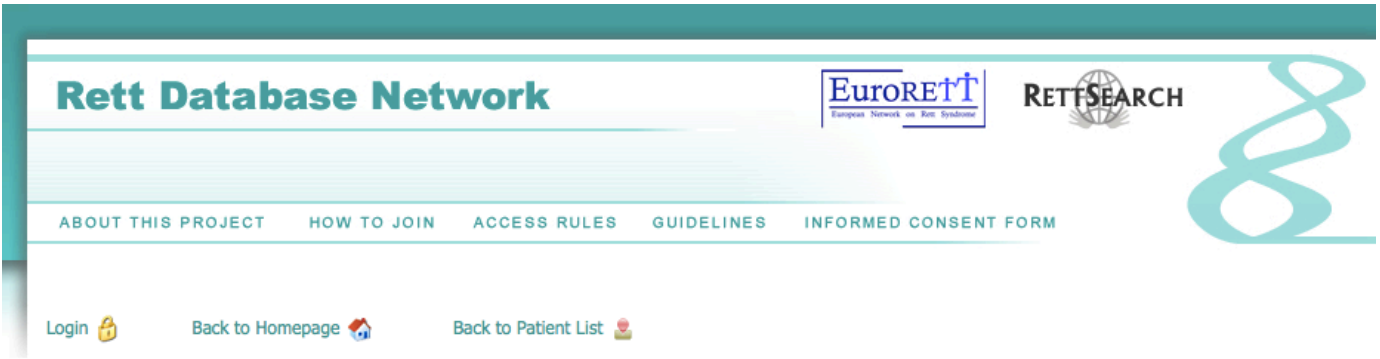
AIM 5: To promote research into Rett syndrome



Number of patients in archive: 2046 (2020 last year)

Australia 1	France 252	Italy 662	Serbia 51
Croatia 29	Germany 0	Poland 0	Spain 426
Czech Republic 0	Hungary 82	Portugal 0	Sweden 0
Denmark 64	India 3	Romania 16	United Kingdom 255
Finland 0	Israel 93	Russia 16	USA 96

http://www.rettdatabasenetwork.org

Pedro Rocha (Catalonia) official database board member reports to RSE

«The aim of this project is to connect the already existing databases and to create a unified repository […] The data will be accessible to the participants and to the scientific community according to rules that assure transparency and equity […] This international effort will be of great value in order to perform genotype-phenotype correlations, to study modifier genes, and to select subgroups of patients for clinical trials.»

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AIM 5: To promote research into Rett syndrome

- FP7 (2007-2013): Modelling in small populations

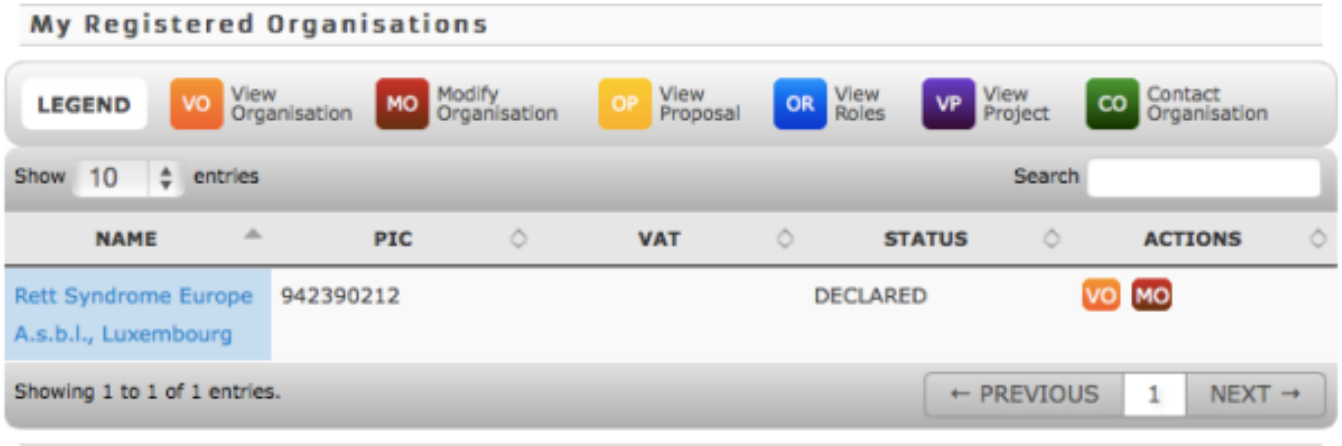
  inSPiRe - *Innovative Methodology for Small Populations Research*

  (Gérard Nguyen – Advisory Board)

  IDeAl - *Integrated Design and Analysis of small population group trials*

  (Gérard Nguyen – Advisory Board)

- H2020 (2014-2020): RSE registered as official organisation



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AIM 5: To promote research into Rett syndrome

1. **<u>Marie Curie ITN (Innovative Training Network): ”SET-NEURO-D” Consortium</u>**
   Project submitted January 2016 – Project not funded

http://ec.europa.eu/programmes/horizon2020/

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Research

AIM 5: To promote research into Rett syndrome

Marie Curie ITN (Innovative Training Network): "SET-NEURO-D" Consortium - SUMMARY

Serotonin is a key neurotransmitter that plays a crucial role in multiple physiological processes in the central nervous system, including cognitive functions such as learning and memory, through the interaction with at least fourteen distinct receptors. Recent breakthrough studies from members of SET-NEURO-D consortium have proposed the serotonin 7 receptor (5- HT7R) subtype as an innovative therapeutic target for treatment of impaired cognitive functions in neurodevelopmental and neurodegenerative diseases.
The consortium will create a Europe-wide partnership through joint efforts aimed to expand the knowledge on the role of 5-HT7R with major emphasis for cognitive impairment in prototypic diseases such as Fragile-X syndrome (FXS), Rett syndrome (RTT), Alzheimer's disease (AD), and Parkinson's disease (PD). SET-NEURO-D integrated research activities will advance the development of new therapeutic options, thus responding to EU recommendation to establish and implement plans to combat rare diseases (FXS, RTT) and AD.
The consortium includes 9 academic research groups and 3 companies from 8 European countries (Italy, Spain, France, United Kingdom, Germany, Austria, Poland, Denmark). The partners are involved in drug target validation and drug discovery and are internationally-recognized experts in the field of 5-HT7R research in various disciplines: medicinal chemistry, cellular and developmental biology, molecular biology, molecular pharmacology, molecular imaging, neurophysiology, and behavioural neuropharmacology. The multidisciplinary and intersectorial environment of SET-NEURO-D will offer high quality training to 12 early-stage researchers that will be enrolled in PhD programs. They will be educated in scientific and transferable skills and enriched with experience in non-academic sector to qualify for career opportunities in academia and private sector. The research-through-training PhD projects accomplish the H2020 strategy for a modern system of education.

Project not funded

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AIM 5: To promote research into Rett syndrome

- H2020 (2014-2020): ERNs – European Reference Network
  - May this year (2016) **Yvonne Milne** was elected by EURORDIS members to be an ePAG (European Patient Advocacy group) representative to serve on the Board of the proposed ERN for the disease group "Rare Congenital Malformations and Intellectual Disability"

Courtesy of Eurordis

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AIM 5: To promote research into Rett syndrome

The principle of ERNs:
- To facilitate access to better and safer healthcare for EU citizens, including cross border healthcare
- To increase access to medical expertise and information for rare conditions
- To facilitate application of results of research
- To develop tools for the improvement of healthcare quality and patient safety

ERN "Rare Congenital Malformations and Intellectual Disability" Dr Jill Clayton-Smith Manchester (UK):
We seek to provide a patient centred network which will meet the needs of those with rare congenital malformation and intellectual disability syndromes, both diagnosed and undiagnosed and will provide an infrastructure for diagnosis, evidence-based management and collection of patient data. The network will add value to existing services, disseminate best practice guidelines to optimise patient care, facilitate training and capacity building in field, inform the work of service commissioners and facilitate research and development of future therapies.

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