5. European Rett Syndrome Congress

02.-04. November 2017 | Berlin

TO CONNECT PEOPLE – TO SHARE KNOWLEDGE

2016-2017 RSE Activity Report

rse

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RSE Board members (2016-2017)

Caroline Lietaer

Yvonne Milne

Danijela Szili

Wilfried Asthalter

Thomas Bertrand

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 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 UIMP "The actual situation and tendencies in Research" (Santander – July 2017)

In the beautiful city of Santander, in the historical Palace of Magdalena, a conference on Rett Syndrome took place, with scientific updates.
An aim of the conference was to refocus the fundraising activities in Spain to concentrate on research, by creating a common fund between Spanish and Catalan associations called FINRETT.
Families on site and abroad (via streaming) gathered with doctors, researchers, directors of medical centres, directors of universities, sponsors and officials from the Ministry of Health.

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

Caroline Lietaer– New RSE Flyer

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

Download pdf file:
http://www.rettsyndrome.eu/wp-content/uploads/2016/12/Rett-Europe-Flyer-Repro.pdf

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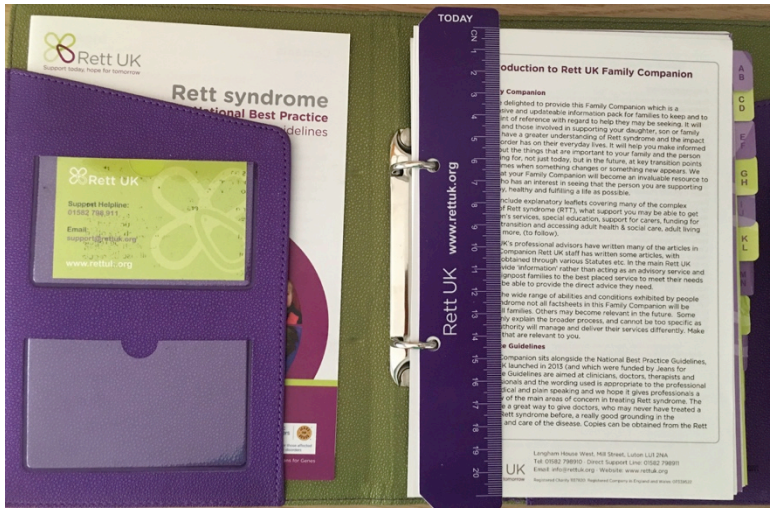
Aim 1: To make Rett syndrome better known to the public, professionals, carers […]

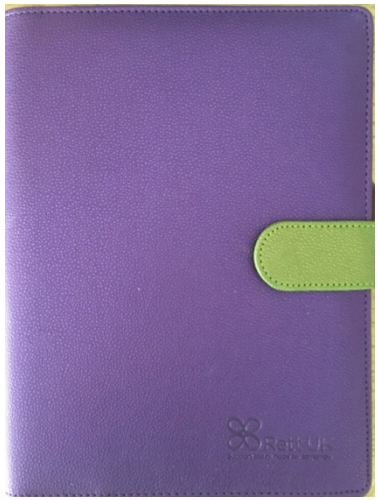
Caroline Lietaer: "Rett Resource", Becky Jenner (RettUK)

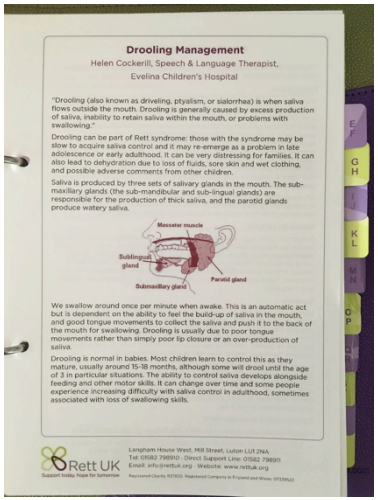
RSE currently has a project underway called the Rett Resource. It aims to gather together information and guidance on a number of important topics relating to the care and management of Rett people. It is based on the framework of UK's Family Companion, but differs from it, as it gathers together international knowledge.

It aims to give doctors and families minimum information, hints about the disease and what to do depending on the situation.
It 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 and Rett Centers.







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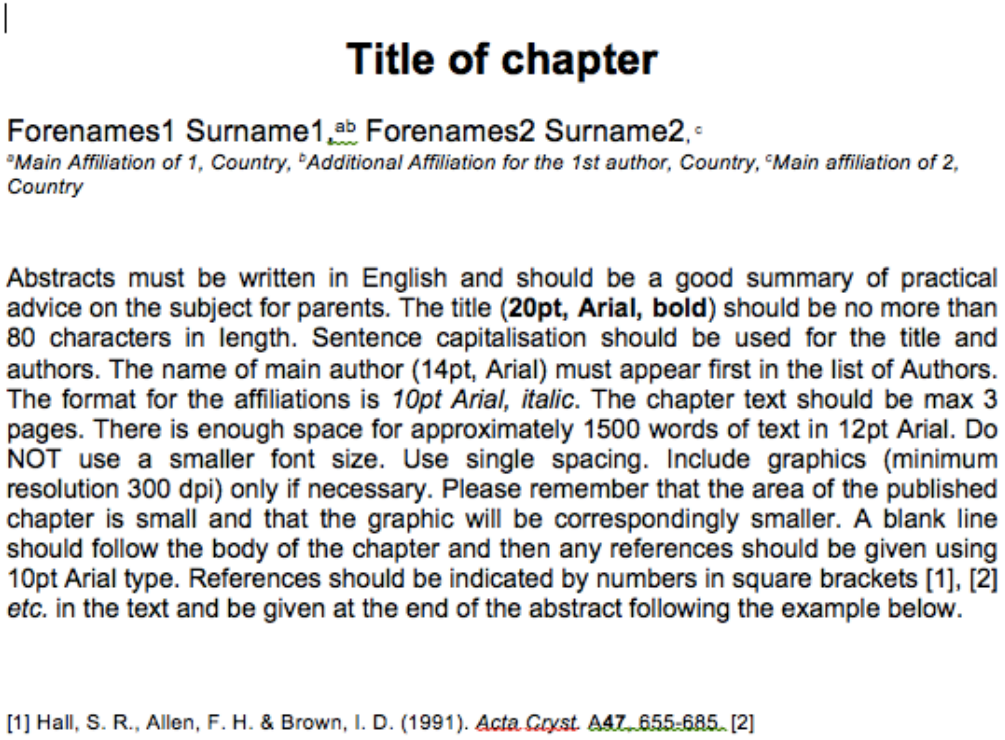
Aim 1: To make Rett syndrome better known to the public, professionals, carers […]

Caroline Lietaer: "Rett Resource"

The topics:

Aging
Breathing irregulaties/airswallowing
Communication
Constipation
Dental care
Depression
Drooling
Epilepsy
Genetics
Hand movement
Hippotherapy
Hydrotherapy
Music therapy
Nutrition
Osteoporosis
Physiotherapy
Puberty
Reflux
Scoliosis
Screaming
Sleeping problems
Toilet training



Title of chapter

Forenames1 Surname1,[ab] Forenames2 Surname2,[c]

[a]Main Affiliation of 1, Country, [b]Additional Affiliation for the 1st author, Country, [c]Main affiliation of 2, Country

Abstracts must be written in English and should be a good summary of practical advice on the subject for parents. The title (**20pt, Arial, bold**) should be no more than 80 characters in length. Sentence capitalisation should be used for the title and authors. The name of main author (14pt, Arial) must appear first in the list of Authors. The format for the affiliations is *10pt Arial, italic*. The chapter text should be max 3 pages. There is enough space for approximately 1500 words of text in 12pt Arial. Do NOT use a smaller font size. Use single spacing. Include graphics (minimum resolution 300 dpi) only if necessary. Please remember that the area of the published chapter is small and that the graphic will be correspondingly smaller. A blank line should follow the body of the chapter and then any references should be given using 10pt Arial type. References should be indicated by numbers in square brackets [1], [2] *etc.* in the text and be given at the end of the abstract following the example below.

[1] Hall, S. R., Allen, F. H. & Brown, I. D. (1991). *Acta Cryst.* **A47**, 655-685. [2]

11 full texts received – proof-reading ongoing
Electronic version prioritized
4000€ of funds have been received

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AIM 2: To improve communication within the European Rett Community

2016-2017 figures:

- Emails

> 250 (e.g. Thomas)

- Rettsyndrome.eu

13 articles, >1000 visitors

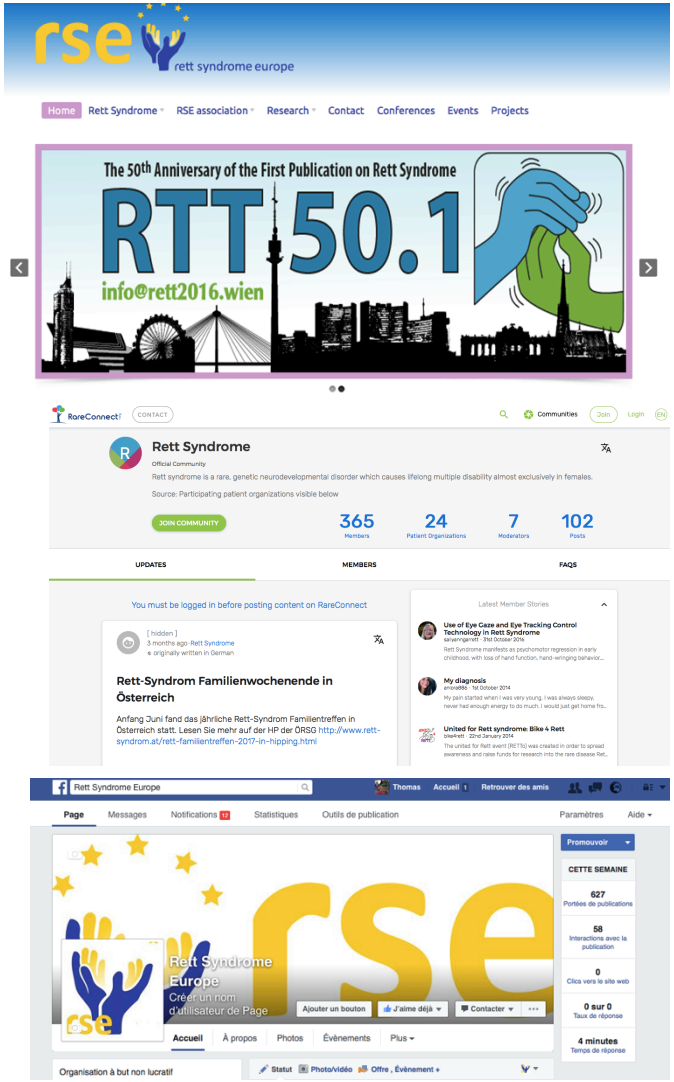
- RareConnect.org

3 posts (only), 365 members, 24 patient organizations, 7 moderators, 102 posts total since creation

Totally re-designed + 8 languages support (English, Spanish, German, French, Italian, Portuguese, Russian and Serbo-Croatian)

- Facebook

public and private groups ~400 average views per post – peak November 2016 (RTT50.1 Vienna, Austria) and June 2017 (Rett University in Krakow, Poland) with >1K views



AIM 2: To improve 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)

Vienna
2016

Berlin
2017

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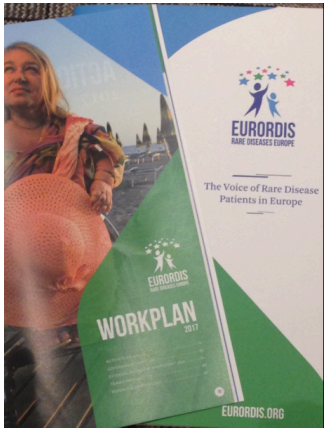
AIM 3: To promote as a representative European organisation, the interests of people with RTT and families

- External Influence and Advocacy

→ *Having RSE officially in the network of European institutions to raise awareness on Rett Syndrome*

1. **RSE is a member of EURORDIS:** Allows RSE to vote at the GAM of EURORDIS During the ECRD (European Congress for Rare Diseases and Orphan products), *Danijela in Budapest (May 2017)*



EURORDIS
RARE DISEASES EUROPE

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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. None from RSE member applied in 2017.

APPLICATION FOR 2018 SUMMER SCHOOL STARTED SEPTEMBER 2017

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External Influence and Advocacy

3. EURORDIS Task Force:

- DITA Drug Information and Transparency Access Task Force (*Danijela Szili)*
  - *Group of patients or patients' representatives giving opinions in the areas of product information, transparency of the regulatory process and access to medicines*
  - *F2F Meeting in November 2015, Danijela re-elected for 3 more years. Friðrik Friðriksson (Iceland) was elected DITA member. Last meeting in Paris (June 2017).*
  - *Recommendations from DITA are sent to the European Medicine Agency (EMA)*

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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HORIZON 2020
The EU Framework Programme for Research and Innovation

European Reference Network
for rare or low prevalence complex diseases

Network Intellectual Disability and Congenital Malformations (ERN ITHACA)

H2020 (2014-2020): ERNs – European Reference Network

May 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"

ITHACA

Intellectual disability, TeleHealth And Congenital Anomalies

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Mission Statement

"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. We will provide an **infrastructure for diagnosis, evidence-based management and collection of secure patient data**. Members of the network will share **best practice** and disseminate **guidelines** to optimise and improve coordination of patient care. We will facilitate **training**, and **capacity building** in field, be active and **collaborative researchers** and work towards development of **diagnostic tests** and **future therapies**."

Prof. Jill Clayton-Smith (University of Manchester, UK)

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Participating Centres

- 38 centres
- 14 member states
- Several centres have expressed an interest
- Awaiting instructions on formal EU process
- In the meantime any centre can be engaged and participate in activities
- Actively seeking other patient members who wish to be involved

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What diseases?

There are many disease represented in ITHACA. Some are bundled into groups, but there are any individual disorders. ITHACA covers:

- Rare chromosome disorders *(< 1 in 2000) includes rare microdeletion syndromes like Koolen-de Vries and Kleefstra
- Chromatin disorders eg Rubinstein-Taybi, De Lange, CHARGE, KAT6B, Kabuki, Coffin Siris, Baraitser Winter, etc
- Neurodevelopmental disorders: Rett, all the Rett variants eg FOXG1 (though EPIRARE covers CDKL5) Angelman, Pitt Hopkins, Mowat Wilson, etc
- Craniofacial disorders, holoprosencephaly, frontonasal dysplasia, clefting disorders, etc
- Rare intellectual disability syndromes, ATRX, Coffin Lowry,
- Overgrowth syndromes, Beckwith, NFIX, Sotos, Weaver, SGB, etc

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What now?

- There are two initiatives where RSE is directly involved:

a) Patient Care/Guidelines – Working on Recent Official French Health Guidelines on Rett Syndrome

b) Patient Registries – H2020 bid from Prof. Alessandra Renieri (Siena, Italy) coordinating an application on ITHACA behalf. This involves harmonisaion of different types of register and concentrates on patient-entered data as a final pathway.

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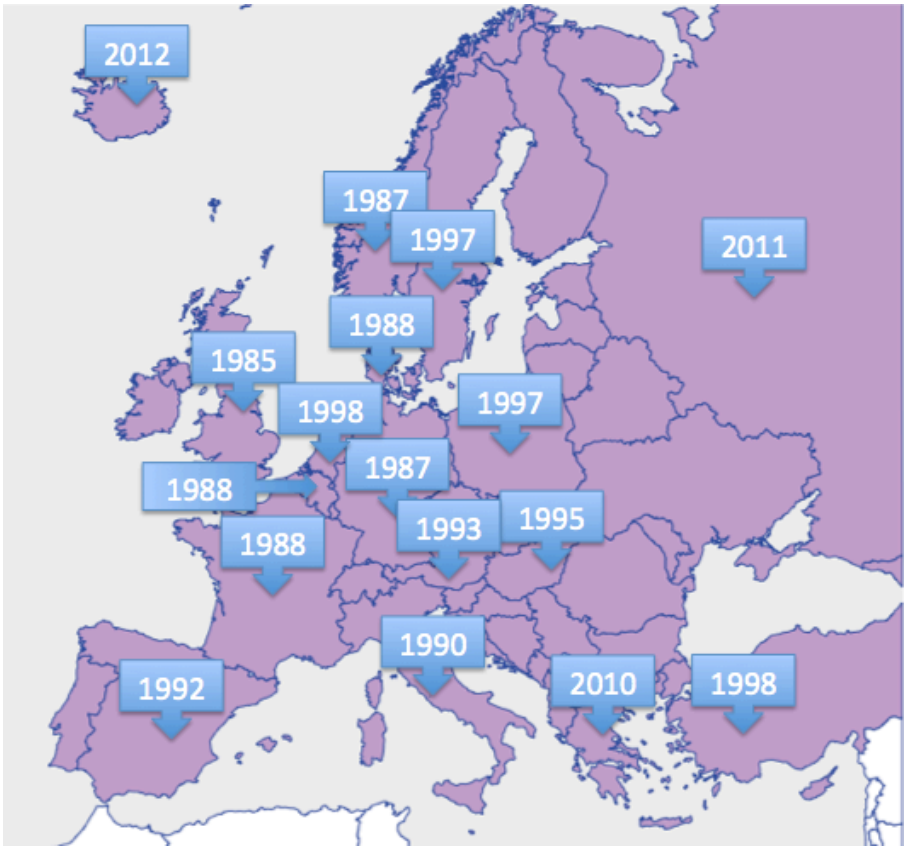
AIM 4: To Expand RSE to all European Countries and to assist, if necessary, in the creation of national associations

→ 42 family associations or family contacts

- ✓ Netherlands
- ✧ Albania?
- ✧ Azerbaijan?
- ✧ Georgia?



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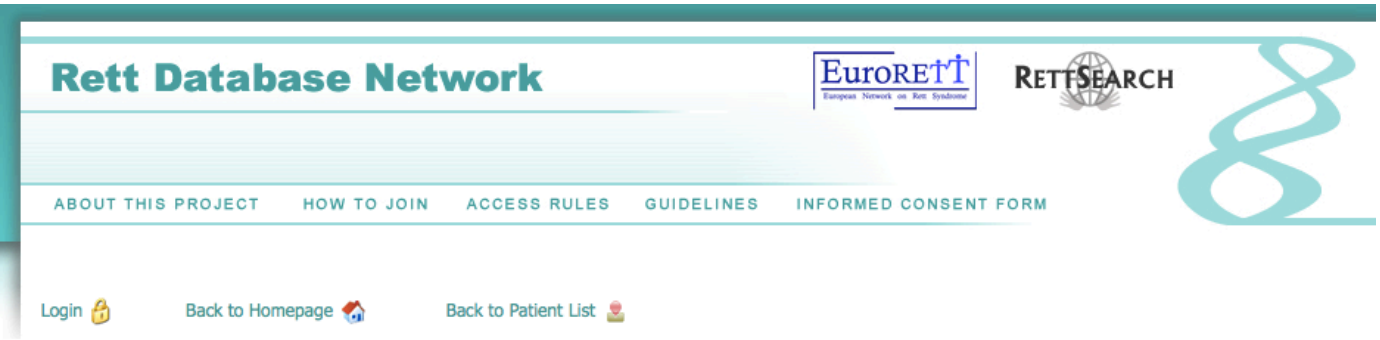
AIM 5: To promote research into Rett syndrome



Number of patients in archive: 2050 (2046 last year)

Australia 1	France 252	Italy 665	Serbia 51
Croatia 29	Germany 0	Poland 0	Spain 427
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

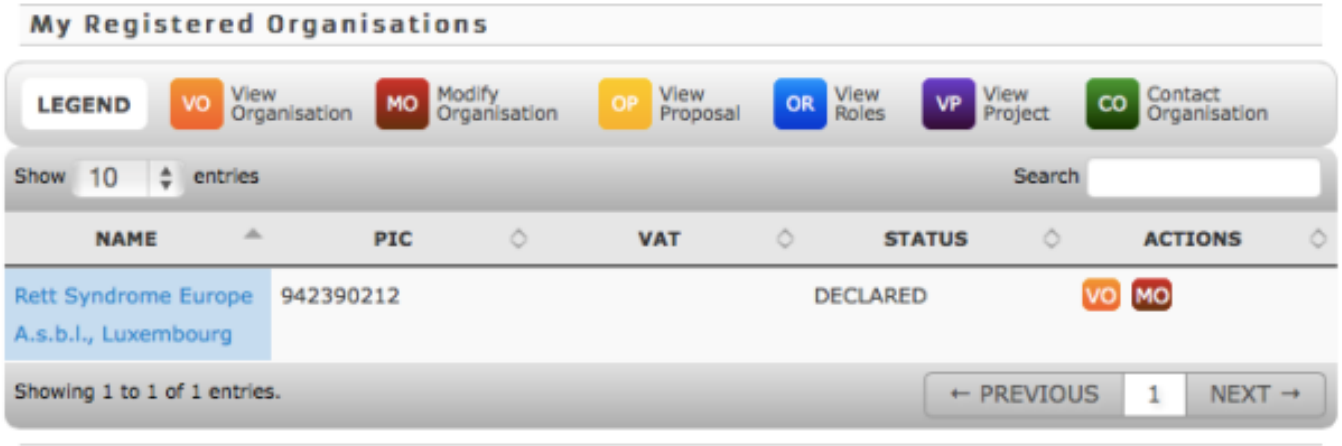
  inSPiRe - *Innovative Methodology for Small Populations Research*

  (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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A.O.B

1. Changes in the RSE statutes
    - Two major changes:

"**Article 10 - Executive Board**

1. The Executive Board (Board of administration) is elected from the Members of the General Assembly and consists of a President, Secretary, Treasurer and two ordinary Members. At least four Members *(was two Members)* of the Executive Board must be family or carers of a person with Rett syndrome. A minimum of three States must be represented on the board.

2. The term for each Executive Board member shall be three years *(was two years)*. They may be elected for a further two terms.

    - Minor changes:
        - Name and addresses of board members
        - Address of registered office
        - Article numbers
        - Dates of amendment laws