

Rett Syndrome Europe Activity Report

2014 - 2015

RSE Board members (2015)



Martine Gaudy



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 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

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

1. Revitalise the network

Constant update of all the contacts in all countries to be able to communicate

2. Inform, advertise special events

News/Articles/Events on RSE website

Building the Network

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

5th Catalan Rett day (Barcelona – April 2015)

This recent event, which took place on April 11 in Barcelona, focused on technological and pedagogical advances in new communication tools for Rett environment

During one day, a wide range of speakers, led by the Catalan deputy director of Education, deputy of Catalan parliament, doctors, researchers, patients' associations, social workers, gave presentations

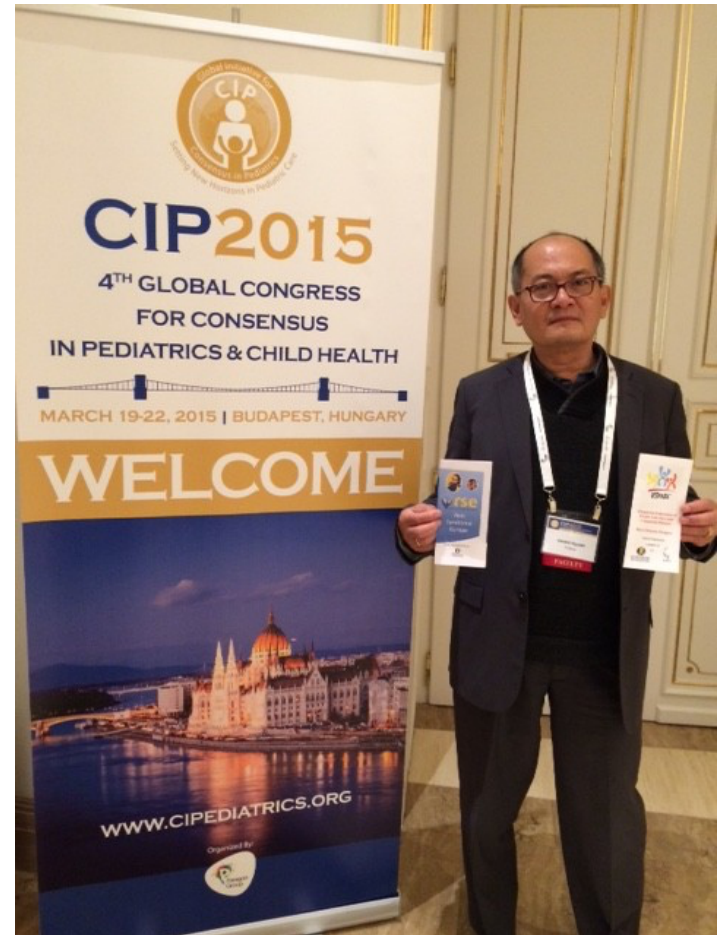


To make Rett syndrome better known to the public, professionals, carers [...]

- ***4th global congress for Consensus in Pediatrics & Child health (Budapest – March 2015)***

Dr Gérard Nguyen (representing both RSE and Hungarian Rett syndrome association) gave a talk entitled:

“Rare Diseases: From Best Care to Innovative Cure”



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

1. Press conference on the presentation of the next World Congress on Rett syndrome
(Thomas Bertrand – January 2015)
 - On January 30, 2015 in the Civic Chamber of the Russian Federation (Moscow) on the initiative of the Civic Chamber of the Republic of Tatarstan and the Association for Assistance to Rett syndrome patients the following events were held:
 - **1. Press-conference on presentation of the World Congress on Rett syndrome in Russia**
 - **2. Roundtable on «Medical and social support and quality of life for girls with Rett syndrome» topic**
2. Extended meeting of the Organising committee in Kazan
(Gérard Nguyen – April 2015)



Building the Network

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

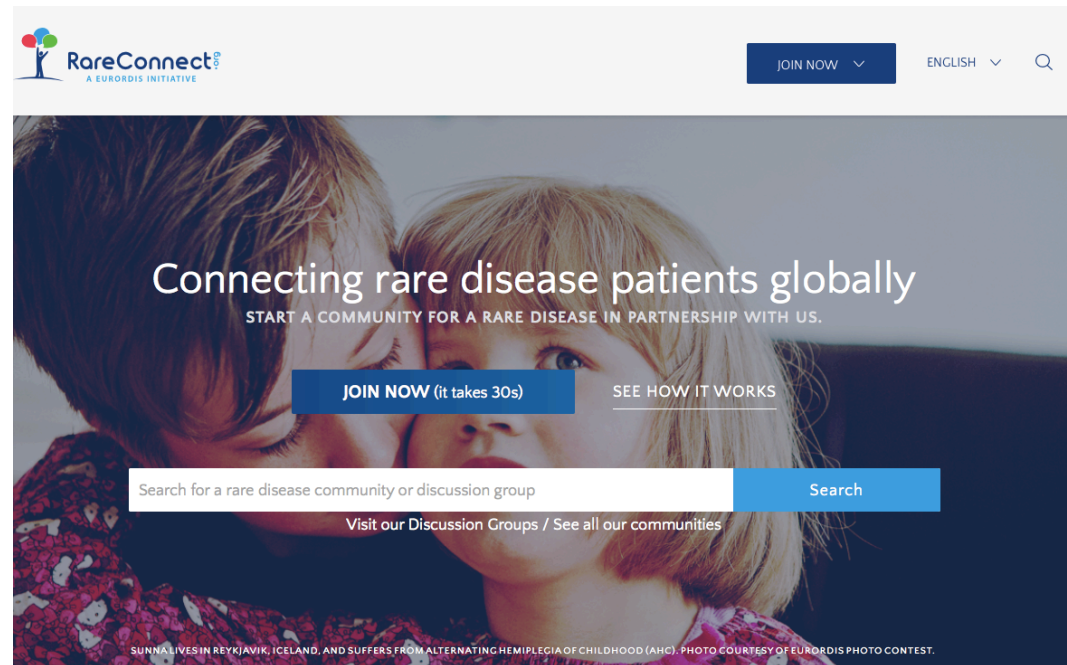
Rettsyndrome.eu

mobile-friendly

RareConnect

Facebook

public and private groups



Building the Network

- **AIM 2:** To improve the communication within the European Rett Community
 - Collaborating in setting up international events on Rett syndrome



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), *Danijela in Madrid (May 2015)*

*Many Rett girls participated in Eurordis Photo contest
Some of the photos were displayed at the ECRD or printed in the Eurordis Activity Report*



the difficult economic and political situation in Greece at the time. For instance, between 2010 and 2014 there were 7 new Ministers of Health. Nevertheless, it was a chance for all stakeholders to get together and to form several partnerships with private health care institutions.

Following considerable was published in July : underway. The legacy o the structures that info first National Plan for R: its implementation. The in place today and is s: EUROPLAN Conference June 2015. This will be implementation of the l

ITALY – UNIAMO, IT FEDERATION FOR R: 11-13 NOVEMBER 2010

The EUROPLAN Confere new model of collaborat institutions as well as w was reproduced in oti different issues include with successful outcom

• 1 – The use of tele gather and share eq by the regional tech optimising resources;

• 2 – Broader neon diseases will be imple

• 3 – Focus on transit needs in all regions ar

LUXEMBURG – AL: LUXEMBURG – 19-20



HUNGARY – HUFERDIS, RARE DISEASES HUNGARY 15-16 OCTOBER 2010 to 25-26 OCTOBER 2013

The EUROPLAN Conference during a period when a new government was being established. The conference was a major milestone in the recognition of rare diseases; it raised awareness amongst the general public through good media coverage. In addition,

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.”

June 2015, Barcelona *DEADLINE FOR 2016 SUMMER SCHOOL IS DECEMBER*

3. EURORDIS Task Force:

- DITA Drug Information and Transparency Access Task Force (*Danijela Szili*)

4. EMA European Medicine Agency (London):

- Scientific Advisory Groups meeting – July 2015 (*Danijela Szili, Friðrik Friðriksson*)

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.

Our Network

- **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?

→ 44 family associations or family contacts

Research

AIM 5: To promote research into Rett syndrome

The screenshot shows the top navigation bar of the Rett Database Network website. It includes the title "Rett Database Network" on the left, and logos for "EuroRETT" (European Network of Rett Syndrome) and "RETTSEARCH" on the right. Below the title are several menu items: "ABOUT THIS PROJECT", "HOW TO JOIN", "ACCESS RULES", "GUIDELINES", and "INFORMED CONSENT FORM". At the bottom of the navigation bar are links for "Login", "Back to Homepage", and "Back to Patient List".



Number of patients in archive: 2012 (2020)

Australia 1	France 252	Italy 654	Serbia 50
Croatia 29	Germany 0	Poland 0	Spain 424
Czech Republic 0	Hungary 58	Portugal 0	Sweden 0
Denmark 64	India 3	Romania 16	United Kingdom 255
Finland 0	Israel 93	Russia 16	USA 96

<http://www.rett databasenet network.org>

Oliviero Dell'Oro (Italy)
Jordi Serra (Catalonia)

«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.»

Research

AIM 5: To promote research into Rett syndrome



1. **Marie Curie ITN (Innovative Training Network): "Networked" for Rett (RETT-net)**
Project not funded
2. **MECP2-related Disorders Consortium – MEDIC**
Project not funded

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

Research

AIM 5: To promote research into Rett syndrome

MECP2-related Disorders Consortium – MEDIC (H2020 PHC14 « New Therapies for Rare Diseases »)

1.1 Objectives

The overall aim of the MEDIC proposal is **to identify exploitable aspects of pathophysiology in MeCP2-disorders and to identify robust biomarkers and novel therapeutic solutions**. To this end, a number of specific objectives need to be met:

1. To develop robust platforms to understand the pathophysiology of MeCP2 disorders, including accurate mouse models for physiology/behaviour and patient-derived cell systems.
2. To identify useful biomarkers of onset and severity during both the prodromal and symptomatic phases in each of the MeCP2 disorders, and to evaluate their use as outcome measures during disease-modifying therapeutic interventions.
3. To identify key molecular and cellular pathways and mechanisms affected by altered levels of functional MeCP2 and to identify whether pharmacological manipulation can be used to intervene in the relevant pathophysiological processes.
4. To assess whether existing and novel genetic and pharmacological approaches can be applied in appropriate preclinical models to reverse or ameliorate the severity of MeCP2 disorder phenotypes.

Project not funded

Research

AIM 5: To promote research into Rett syndrome

Marie Curie ITN (Innovative Training Network): "Networked" for Rett (RETT-net)

Participants are the major European centers working on Rett syndrome in Italy (Siena), France, UK (Cardiff), Israel, Hungary and Spain.

The aim of the program is the activity in the field of Rett syndrome to foster exchanges of relevant experience, policies and practices between European countries including:

- Enhancing the visibility and recognition of the international registry, Rett Networked Database and improve accuracy of data
- Connecting the Rett Networked Database to biobank resources
- Contributing to the development and dissemination of knowledge on Rett syndrome through to the support of patients' association
- Contributing to improvements in access to quality services from diagnosis to care

The plan is to recruit researchers for filling the Rett networked database, improve the biobanks and offering them a training in Rett (at least 7 for a period of 3 years) in a way that each center has at least 1 MD per year. Each center will have 1800 Euros per month in order to plan formation (the recruitment of researchers to be trained; training and networking costs, organization of joint activities and conferences; management and overheads).







Project not funded

Research



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

My Registered Organisations

LEGEND  View Organisation  Modify Organisation  View Proposal  View Roles  View Project  Contact Organisation

Show entries

NAME	PIC	VAT	STATUS	ACTIONS
Rett Syndrome Europe A.s.b.l., Luxembourg	942390212		DECLARED	 

Showing 1 to 1 of 1 entries.