



# 4TH EUROPEAN CONGRESS ON RETT SYNDROME

Researchers and Clinicians Program, with CME credits  
for Physicians, Biologists and Geneticists

 **AIRETT**  
*da 25anni ricerca, cuore, cura*

- 1 – Researchers and Clinicians Program, with CME credits for Physicians, Biologists and Geneticists
- 2 – Therapists, Educators and Teachers Program, with different CME credits for Therapists and Educators

Exhibition venues for aids, equipments and other medical devices

Individual counseling for families about specific issues

Families are admitted at all the sessions

Workshop on the SUD in Rett Syndrome for clinicians of the Committee and representatives of Associations

## RESEARCHERS AND CLINICIANS PROGRAM PLENARY SESSION

**Friday, October 30th**

**9.00**

### Opening Ceremony

Dovigo L (AIRett Rome, Italy) - Bertrand T (RSE Paris, France) - Nues P (Rettsyndrome.org, Danville, USA)

Welcome Remarks

**9.20**

### Updates on the European policy about Rare Diseases

Taruscio D (ISS Rome, Italy)

Moderator: Veneselli E (G Gaslini Institute, University of Genoa, Italy)

**9.40**

### Basic Research 1

9.40 Gabel HW (Washington University Medical School, USA)

Disruption of DNA-methylation-dependent long gene repression

10.15 Landsberger N (San Raffaele Scientific Inst., University of Milan, Italy)

Studying MeCP2 phosphorylation to obtain novel insights on MeCP2 functions and Rett syndrome

10.40 2 selected oral abstract presentations

**11.00 Discussion**

Moderators: D'Esposito M (IGB Napoli, Italy) - Ballas N (Stony Brook University, New York, USA)

**11.15**

Discussion

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**11.45**

**Basic Research 2**

**11.45 Heidenreich M (Broad Institute of MIT and Harvard, USA)**

In vivo interrogation of MeCP2 using CRISPR-Cas9

**12.10 Ballas N (Stony Brook University, New York, USA)**

Glial cells, MECP2 and Rett Syndrome

**12.35 Huppke P (University of Gottingen, Germany)**

Wild-type microglia does not reverse pathology in mouse models of Rett syndrome

**13.00 Discussion**

*Moderators: Gabel HW (Washington University Medical School, USA) - Pizzorusso T (University of Firenze, Italy)*

**13.15**

**Lunch Break & Poster Viewing**

**14.30**

**From an idea to a pharmaceutical product**

**Garattini S (IRCCS Istituto di Ricerche Farmacologiche Mario Negri, Milan, Italy)**

From an idea to a pharmaceutical product

**14.50**

**Evolution over time and over the lifespan**

**Leonard H (Telethon Kids Institute, Perth, Australia)**

*Moderator: Curatolo P (University of Rome Tor Vergata, Italy)*

**15.10**

**Evolution and Treatment from Childhood to Adulthood**

**15.10 Pineda M (Hospital Sant Joan de Déu, Barcelona, Spain)**

Protocols for Transitioning care from Childhood to Adulthood

**15.30 Wilken B (Klinikum Kassel, Germany) - Management of Transition**

Management of Transition

**15.50 Discussion**

*Moderator: Budden S (Oregon, USA) - Zappella M (University of Siena, Italy)*

**16.10**

**Coffee Break**

**16.30**

**What we have learned about Epilepsy and Sleep**

**16.30 Glaze D (Blue Bird Circle Rett Center and Texas Children's Sleep Center, Texas, USA)**

*Moderator: Vigeveno F (Ospedale Pediatrico Bambino Gesù, Rome, Italy)*

**17.10**

**Focus on Epilepsy**

**17.10 Nissenkorn A (University of Tel Aviv, Israel)**

Updates on Epilepsy

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**17.30 Cusmai R (Ospedale Pediatrico Bambino Gesù, Rome, Italy)**

Epileptic and non-epileptic paroxysmal events

**17.50 Vignoli A (University of Milan, Italy)**

Efficacy of new and alternative treatments

**18.10 Discussion**

*Moderators: Vigevano F (Ospedale Pediatrico Bambino Gesù, Rome, Italy) - Pineda M (Hospital Sant Joan de Déu, Barcelona, Spain)*

**18.30**

**Genetics**

**18.30 Russo S (Auxologico Inst., Milan, Italy)**

Exome sequencing of a cohort of Rett patients: new involved genes

**18.50 Armstrong J (Hospital Sant Joan de Déu, Barcelona, Spain)**

Improving genetic diagnosis using NGS: comparing exomes panels

**19.10 1 selected oral abstract presentation**

**19.20 - 19.30 Discussion**

*Moderators: Clarke A (Institute of Cancer & Genetics, Cardiff University, UK) - Renieri A (University of Siena, Italy)*

**Saturday, October 31st**

**9.00**

**Pharmacological and Genetic Approaches 1**

**9.00 Green M (University of Massachusetts Medical School, USA)**

Genetic and Pharmacological Reactivation of the Mammalian Inactive X Chromosome: Implications for Treatment

**9.25 Cobb S (University of Glasgow, UK)**

Gene Therapy

**9.50 Discussion**

*Moderators: Villard L (Marseille Medical School, Faculté de Médecine de La Timone, Marseille, France) - Laviola G (ISS, Rome, Italy)*

**10.00**

**Pharmacological and Genetic Approaches 2**

**10.00 Ciani E (University of Bologna, Italy)**

Therapies for CDKL5 disorder

**10.25 Giustetto M (University of Turin, Italy)**

Synaptic and behavioural phenotypes in MECP2 and CDKL5 mouse models: insights into pharmacological therapies

**10.50 Discussion**

*Moderators: Huppke P (University of Gottingen, Germany) - Hayek J (University of Siena, Italy)*

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**11.00**  
**Coffee Break**

**11.30**  
**Pharmacological and Genetic Approaches 3**  
4 selected oral abstract presentations

**12.10 Discussion**

*Moderators: Huppke P (University of Gottingen, Germany) - Hayek J (Policlinico Le Scotte, Siena, Italy)*

**12.20**  
**Assistance Advances and Complex Issues: Care and Prevention 1**

**12.20 Budden S (Oregon, USA)**  
Recognition and Management of Autonomic Disturbance

**12.40 Veneselli E (G Gaslini Institute, University of Genoa, Italy)**  
Improvements in Emergency Assistance: the Clinical Passport

**13.00 Discussion**

*Moderators: Canevini M.P (University of Milan, Italy)*

**13.10**  
**Lunch Break & Poster Viewing | General Assembly RSE**

**14.30**  
**Assistance Advances and Complex Issues: Care and Prevention 2**

**14.30 Kendrick A (University Hospitals, Bristol, UK)**  
Breathing problems and their diagnosis

**14.50 Chiarini Testa MB (Ospedale Pediatrico Bambino Gesù, Rome, Italy)**  
Breathing problems and their treatment (management)

**15.10 Discussion**

**15.20 Canevini MP (University of Milan, Italy)**  
The management of epilepsy in complex disabilities

**15.40 Rimini A (G Gaslini Institute, University of Genoa, Italy), Mancardi MM (G Gaslini Institute, University of Genoa, Italy)**  
Preventive Cardiology (cardiological prevention) and drug interactions

**16.00 Cefalo G (University of Milan, Italy)**  
The management of nutrition related to age and disability

**16.20 Discussion**

*Moderators: Wilken B (Klinikum Kassel, Germany), Papoff P. (La Sapienza University, Rome)*

**16.30**  
**Coffee Break**

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**17.00**

**New Insights in Cognition and Communication**

**17.00 Djukic A (Montefiore Medical Center, New York, USA)**

Communication and cognitive empowerment

**17.20 Fabio RA (University of Messina, Italy)**

On the bridge between neurological and psychological factors: TdCS and cognitive empowerment

**17.40 Discussion**

*Moderators: Leonard H (Telethon Kids Institute, Perth, Australia) - Leuzzi V. (La Sapienza University of Rome, Italy)*

**18.00**

**Movement Disorders: Evaluation and Rehabilitation**

**18.00 Leonard H (Telethon Kids Institute, Perth, Australia)**

Motor development: new frontiers for walking activity evaluation

**18.20 Lotan M (Ariel University, Israel)**

Rehabilitation: advances and new trials

**18.40 - 19.00 Discussion**

*Moderators: Budden S (Oregon, USA) - Rodocanachi M (Fondazione Don Carlo Gnocchi, Milan, Italy)*

**Sunday, November 1st**

**9.00**

**Focus on Clinical Trials: From Scientific Background to Results; How to Change the Therapies**

**9.00 Kaufmann W (Harvard Med. School, USA)**

The lessons from IGF-1 trials

**9.20 Glaze D (Blue Bird Circle Rett Center and Texas Children's Sleep Center, Texas, USA)**

NNZ-2566: a novel experimental treatment

**9.40 Djukic A (Montefiore Medical Center, New York, USA)**

Pharmacological treatments with Copaxone and Statins

**10.00 Ben Zeev B (Sourasky Medical Center, Tel Aviv, Israel)**

Copaxone from cells to RTT patients -hopes and unexpected adverse events

**10.20 Discussion**

**10.40 Coffee Break**

**11.00 Mancini J. (Hôpital de la Timone, Marseille, France)**

Desipramine and cardio-respiratory variability

**11.20 Rothenbuhler A. (Hôpital Bicêtre, Paris, France)**

Bone Health in Rett Syndrome

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## 11.40 Hayek J. (Policlinico Le Scotte, Siena, Italy)

EPI-743 trial results

## 12.00 Discussion

Moderators: Villard L (Marseille Medical School, Faculté de Médecine de La Timone, Marseille, France) Clarke A (Institute of Cancer & Genetics, Cardiff University, UK)

## 12.10

### Perspectives on the future

## 12.10 Nguyen G (Hôpital Avicenne – Hôpitaux Universitaires Paris-Seine-Saint-Denis, France)

Rethinking the future of clinical trials in Rett Syndrome

## 12.25 Mari F (University of Siena, Italy)

Updates on Rett Networked Database

## 12.40 De Montleau B (Eurordis)

Supporting research in Europe

## 12.55 Discussion

Moderators: Leonard H (Telethon Kids Institute, Perth, Australia), Hayek J (Policlinico Le Scotte, Siena, Italy)

## 13.10

### Final Meeting and Experts Conclusions

Research experts final considerations

Clinic experts final considerations

Rehabilitation experts final considerations

## 13.30

Closing remarks

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