

# RETT SYNDROME RESEARCH, TOWARDS THE FUTURE

ROME  
27-29 September 2018

The aim of this scientific meeting is to provide a platform to discuss **recent advances** in basic, translational and clinical research on **Retts syndrome**.

*The meeting is also intended to promote and foster collaborations.*

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## RETT SYNDROME RESEARCH, TOWARDS THE FUTURE

### Program

27 September, 2018

08,45 - 10,45: REGISTRATION

10,45 - 11,00 WELCOME AND OPENING REMARKS

**Session 1: Basic research on MeCP2: from its structural domains to its essential molecular functions**

Chair: **James Eubanks**

11:00 - 11:40

**Adrian Bird**, University of Edinburgh (UK)  
*The molecular basis of Rett syndrome*

11:40 - 12:10

**Juan Ausio**, University of Victoria (Canada)  
*The two isoforms of MeCP2 in search of a function*

12:10 - 12:30

**Francesco Bedogni**, San Raffaele Scientific Institute (Italy)  
*Lack of Methyl-CpG binding protein 2 (MeCP2) affects cell fate refinement during embryonic cortical development*

12:30 - 12:45

**Nicolas Panayotis**, Weizmann Institute of Science (Israel)  
*Importin alpha5 is a new in vivo target for modulation of MeCP2 pathways*

12:45 - 13:00

**Alessio Cortelazzo**, Azienda Ospedaliera Universitaria Senese (Italy)  
*Brain protein changes in Mecp2 mouse mutant models: effects on disease progression of Mecp2 brain specific gene reactivation*

13:00 - 14:30 Lunch break

**Session 2: Latest news from the development of novel cellular and animal models in Rett research**

Chair: **Juan Ausio**

14:30 - 15:10

**Zhaolan Zhou**, University of Pennsylvania (USA)  
*"Seq-ing" pathogenic insights into Rett syndrome*

15:10 - 15:40

**Nicoletta Landsberger**, University of Milan and San Raffaele Scientific Institute (Italy)  
*Is precision medicine relevant for the treatment of Rett syndrome? Suggestions from a novel mouse model of Mecp2*

15:40 - 16:00

**Diego Sbardella**, University of Rome (Italy)  
*Dys-regulation of autophagy and proteasome leads to altered proteostasis in Rett syndrome*

16:00 - 16:20

**Claudia Fuchs**, University of Bologna (Italy)  
*Alteration of SMAD3 signaling decreases hippocampal neuronal survival in Cdkl5 KO mice*

16:20 - 16:40 Coffee break

**Session 3: New insights in neuronal and non-neuronal dysfunctions generated by MeCP2 deficiency**

Chair: **Enrico Tongiorgi**

16:40 - 17:20

**Jeffrey Lorenz Neul**, Vanderbilt University Medical Center (USA)  
*Autonomic dysfunctions and approaches to therapy in Rett syndrome*

17:20 - 17:50

**Lucas Pozzo-Miller**, The University of Alabama at Birmingham (USA)  
*Dysfunction of the hippocampus-prefrontal cortex projection and its role in social deficits in Rett syndrome*

17:50 - 18:10

**Maurizio Giustetto**, University of Turin (Italy)  
*Loss of Mecp2 causes atypical synaptic and molecular plasticity of parvalbumin-expressing interneurons reflecting Rett syndrome-like sensorimotor defects*

18:10 - 18:30

**James Eubanks**, Krembil Research Institute Toronto (Canada)  
*The surveillance receptor TRPM2: a novel therapeutic target for Rett syndrome?*

18:30 - 18:50

**Flavia Antonucci**, University of Milan (Italy)  
*Tuning of ATM activity to control the delayed GABA development and neuronal hyper-excitability in neurodevelopmental disorders*

19:00 - 20:30 Finger food and poster session

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**Session 4: Treating Rett syndrome: future approaches and novel challenges**

Chair: **Nino Ramirez**

8:45 - 9:15

**Stuart Cobb**, University of Edinburgh (UK)  
*An update on gene therapies for Rett syndrome*

9:15 - 9:45

**Joost Gribnau**, Erasmus MC (NL)  
*In vivo and in vitro models for X chromosome reactivation*

9:45 - 10:15

**Antonio Casini**, University of Trento (Italy)  
*From user - to genome-friendly CRISPR/Cas9*

10:15 - 10:35

**Alessandra Renieri**, University of Siena (Italy)  
*Toward gene editing in Rett syndrome*

10:35 - 11:00 Coffee break

## Session 5: What is going on: preclinical studies in MeCP2 related disorders

Chair: **Maurizio Giustetto**

11:00 - 11:30

**Steve Kaminsky**, Rettsyndrome.org (USA)  
*IRSF's Scout Program: an in vivo screen for potential drugs to treat Rett syndrome*

11:30 - 12:00

**Enrico Tongiorgi**, University of Trieste (Italy)  
*Repurposing Mirtazapine to target Rett syndrome*

12:00 - 12:20

**Daniele Vigli**, Istituto Superiore di Sanità (Italy)  
*The phytocannabinoid cannabidiol (CBDV) rescues behavioural alterations and brain atrophy in Mecp2-308 hemizygous male mice*

12:20 - 12:40

**Jean Christophe Roux**, Aix Marseille University (France)  
*Calcineurin-huntingtin pathway restores BDNF trafficking and improves Mecp2 knock-out mice symptoms*

12:40 - 14:15 Lunch break

## Session 6: What is going on: news from ongoing clinical studies and trials

Chair: **Nicoletta Landsberger**

14:15 - 14:55

**Alan K. Percy**, University of Alabama at Birmingham (USA)  
*Rett syndrome: closing the gap to clinical trials*

14:55 - 15:35

**Walter E. Kaufmann**, Emory University & Mind Institute/University of California Davis (USA)  
*Clinical trials in Rett syndrome: a critical appraisal*

15:35 - 16:05

**Ravi Anand**, Newron Pharmaceutical SpA (Italy)  
*Sarizotan in the treatment of respiratory abnormalities in patients with Rett syndrome (RTT): new findings from an international 6-month, randomized, double-blinded, placebo controlled, phase III trial (stars)*

16:05 - 16:30 Coffee break

16:30 - 17:00

**Jan-Marino Ramirez**, Center for Integrative Brain Research Seattle Children's Research Institute and University of Washington (USA) (USA)  
*Dysautonomia in Rett syndrome: from bench to bedside*

17:00 - 17:20

**Breanne Byiers**, University of Minnesota (USA)  
*Behavioral and cardiac reactivity to sensory stimulation in Rett syndrome*

17:20 - 17:40

**Gillian Townend**, Rett Expertise Centre Netherlands (NL)  
*Oculomotor function in individuals with Rett syndrome*

17:40 - 18:00

**Murim Choi**, Seoul National University College of Medicine (PRK)  
*GABBR2 mutations determine phenotype in Rett syndrome and epileptic encephalopathy*

18:30 Short tour of Rome by bus followed by a social dinner in front of the Colosseum (dress code: casual)

## 29 September, 2018

### In traduzione simultanea: La ricerca sulla sindrome di Rett: lo stato attuale e le prospettive future

Moderatori: **Salvatore Franzè, Nicoletta Landsberger**

9:30 - 10:00

**Salvatore Franzè**, Presidente Pro RETT Ricerca  
**Nicoletta Landsberger**, Università Statale di Milano e Istituto Scientifico San Raffaele (Italia)  
*Introduzione e riassunto delle giornate precedenti*

10:00 - 10:25

**Alan K. Percy**, University of Alabama at Birmingham (USA)  
*Conoscenze e lacune da colmare per i prossimi trial clinici*

10:25 - 10:50

**Walter E. Kaufmann**, Emory University & Mind Institute/University of California Davis (USA)  
*Valutazione critica dei trial clinici per la sindrome di Rett*

10:50 - 11:15

**Jeffrey Lorenz Neul**, Vanderbilt University Medical Center (USA)  
*Disfunzioni autonome e approcci terapeutici per la sindrome di Rett*

11:15 - 11:30 Coffee break

11:30 - 11:55

**Ravi Anand**, Newron Pharmaceutical SpA (Italia)  
*Il Sarizotan per il trattamento delle anomalie respiratorie nei pazienti affetti da sindrome di Rett: i risultati ottenuti da un trial clinico internazionale di fase III*

11:55 - 12:20

**Stuart Cobb**, University of Edinburgh (UK)  
*Lo stato attuale delle terapie geniche per la sindrome di Rett*

12:20 - 12:40

**Antonio Casini**, Università di Trento (Italia)  
*Oltre i limiti del CRISPR/Cas9: miglioramento della fedeltà e del "delivery"*

12:40 - 13:45 Lunch break

13:45 - 14:10

**Steve Kaminsky**, Rettsyndrome.org (USA)  
*Il Programma Scout di IRSF - uno screening in vivo alla ricerca di farmaci utili al trattamento della sindrome di Rett*

14:10 - 14:30

**Angelisa Frasca**, Università Statale di Milano (Italia)  
*I benefici di un nuovo approccio terapeutico in un modello sperimentale di sindrome di Rett*

14:30 - 14:50

**Gillian Townend**, Rett Expertise Centre (NL)  
*Linee guida internazionali per la comunicazione con i pazienti affetti da sindrome di Rett*

14:50 - 15:10

**Enrico Tongiorgi**, Università di Trieste (Italia)  
*Il riposizionamento della Mirtazapina per il trattamento della sindrome di Rett*

15:10 - 15:30

**Dennis Dionne**, Newron Pharmaceuticals SpA (Italia)  
*Una vita intrappolata: un faro sul bisogno inascoltato di accrescere la consapevolezza sulla sindrome di Rett e di fornire informazioni alle politiche pubbliche; uno studio internazionale sulla sindrome di Rett e sull'onere della malattia*

15:30 Saluti e chiusura dei lavori



Associazione per la ricerca sulla sindrome di Rett Onlus

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