

4° CONGRESSO EUROPEO SULLA SINDROME DI RETT

ROMA, 30/31 OTT/1 NOV 2015 - Barcelò Aran Mantegna Hotel

Più di 60 interventi

con la presenza di specialisti tra i più importanti nomi a livello internazionale.

SESSIONE PLENARIA

Aggiornamento sullo stato della ricerca e sui trial in corso.

Presenza in carico Clinica con particolare attenzione alle problematiche più frequenti e importanti nella Rett.

SESSIONE PARALLELA

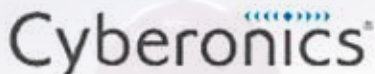
dedicata agli aspetti motori e cognitivi, come intervenire, come affrontarli, come aiutare le ragazze affette da Rett.

Congresso con crediti ECM
AIRETT è riconosciuta dal MIUR
come ente formativo del
personale scolastico.

Con il patrocinio di



In collaborazione con



Da 25 anni
Ricerca, cuore, cura.



Cari tutti,

questo 4° Congresso Europeo è organizzato da AIRett (Associazione Italiana Sindrome di Rett), con la collaborazione di RSE (Associazione Europea Sindrome di Rett), AIRett è stata anche promotrice del 1° Congresso Europeo tenutosi a Milano nel 2009.

In questo interessante evento noi incontreremo insieme ricercatori, clinici, terapisti, educatori, insegnanti, famiglie e le nostre bambine dagli occhi belli.

Lo scopo che ci poniamo è quello di migliorare la nostra assistenza clinica condividendo le nostre competenze ed i nostri studi, nella speranza di nuove idee per un futuro migliore per le ragazze con la Sindrome di Rett (SR).

I ricercatori ci riferiranno i risultati dei loro più recenti studi. Noi sappiamo che stanno lavorando molto duramente per identificare i meccanismi sottostanti la condizione Rett.

Molti di essi hanno testato sugli animali potenziali nuovi trattamenti per far regredire o perlomeno arrestare l'evoluzione della SR. Sarà pertanto molto interessante conoscere i loro riscontri.

Nel contempo, come clinici, noi speriamo di migliorare la presa in carico quotidiana. Quindi i medici più competenti ci parleranno delle loro esperienze per permetterci di curare al meglio le nostre ragazze e di gestire nel migliore dei modi i loro vari problemi clinici, sulla base degli specifici protocolli che ci presenteranno. Abbiamo chiesto loro un focus sul controllo dei sintomi e sulle strategie per prevenire le complicazioni che possono insorgere. Questo punto costituisce un target molto importante nel nostro lavoro quotidiano. Essi ci supporteranno anche nel seguire le ragazze durante l'avanzamento dell'età: i loro suggerimenti indicheranno come farle crescere nelle condizioni migliori possibili e assicurare una qualità di vita confortevole a loro ed ai loro genitori.

Negli ultimi anni, le ragazze con SR hanno partecipato a molti trial clinici con nuove molecole. Abbiamo invitato i principali investigatori per arrivare direttamente a conoscere i risultati ottenuti e discutere la possibilità di trasferire tali esperienze nel nostro lavoro clinico.

Nello stesso tempo abbiamo chiamato gli esperti sulla comunicazione e sulla riabilitazione per ricevere suggerimenti su moderni programmi di trattamento. Li vedremo al lavoro con le ragazze direttamente o tramite videoregistrazioni. Ci saranno presentati inoltre i più nuovi ausili e strumenti.

Noi crediamo nei giovani e li vogliamo con noi per presentarci i risultati dei loro studi.

Infine, abbiamo organizzato un percorso specifico per terapisti, educatori, insegnanti e famiglie in modo che possano confrontarsi e condividere le problematiche con gli esperti, e tornare nelle loro sedi operative con nuove competenze.

Siamo orgogliosi di avere con noi molti dei leaders nei vari campi, non solo dall'Europa ma anche dagli Stati Uniti e dall'Australia, e desideriamo ringraziarli tutti per la loro disponibilità.

Le famiglie potranno partecipare a tutte le sessioni, discutere con gli esperti e avere consulenze individuali con gli esperti stessi.

Vi aspettiamo quindi numerosi a Roma per sfidare la Sindrome di Rett.

Edvige Veneselli – Lucia Dovigo

Since 25 years
Research, heart, care.



Dear all,

this 4th European Congress is organized by AIRETT (Italian Association of Rett Syndrome), with the collaboration of RSE (Rett Syndrome European Association). AIRETT was already the promoter of the first European Congress held in Milan in 2009.

In this interesting event we will meet together, researchers, clinicians, therapists, educators, teachers, families and our girls with beautiful eyes.

The aim is to improve our clinical care sharing our skills and studies, hoping in new ideas for a better future of girls with Rett Syndrome.

The researchers will report the results of their most recent studies. We know that they worked very hard to identify the mechanisms underlying the Rett condition.

Many of them tested potential new treatments on animals in order to reverse or stop the evolution of RS. So it will be very interesting to know their findings.

Meanwhile, as clinicians, we wish to improve daily care. Thus the most skilled practitioners will talk about their experiences allowing us to better care our girls and manage the various clinical problems, on the basis of specific protocols that they will present.

We asked them to focus on symptoms control and on strategies to prevent complications that may arise. This point constitutes a very important target in our daily work. They also support us in following girls during aging: their suggestions will show how to make girls grow in the best possible conditions and ensure a comfortable quality of life for them and their parents.

In recent years, many clinical trials with new molecules have been carried on in girls with Rett Syndrome. We invited principal investigators to directly get aware of their results and discuss about the possibility of transfer their findings to our clinical work.

At the same time we called experts on communication and rehabilitation to receive suggestions about modern treatment programs. We wish to look at them working with girls directly or through videotape.

Moreover they will present the newest aids and tools.

We believe in young people and want them with us to present the findings of their studies.

Finally, we organized a specific track for therapists, educators, teachers, families so that they can compare and share questions with the experts and get back to their homes with new skills.

We are proud to have with us many of the leaders in various fields, not only from Europe but even from the United States and Australia, and we want to thank all of them for their availability.

Families can participate in all sessions, discuss with all the experts and have individual consultations with the experts.

Therefore, we expect many of you in Rome for the challenge to the Rett Syndrome.

Edvige Veneselli – Lucia Dovigo

General Scheme - Schema generale

1 – Researchers and Clinicians Program, with CME credits for Physicians, Biologists, Geneticists, Pharmacists, Biologists, Nurses, Pediatric nurses.

Programma per Ricercatori e Clinici, con crediti ECM per Medici, Biologi, Genetisti, Farmacisti, Biologi, Infermieri, Infermieri pediatrici.

Room A - Aula A.

2 – Therapists, Educators and Teachers Program, with different CME credits.

Programma per Terapisti, Educatori e Insegnanti, con crediti ECM per Fisioterapisti, Terapisti della Neuro e Psicomotricità dell'età evolutiva, Logopedisti, Psicologi, Educatori professionali.

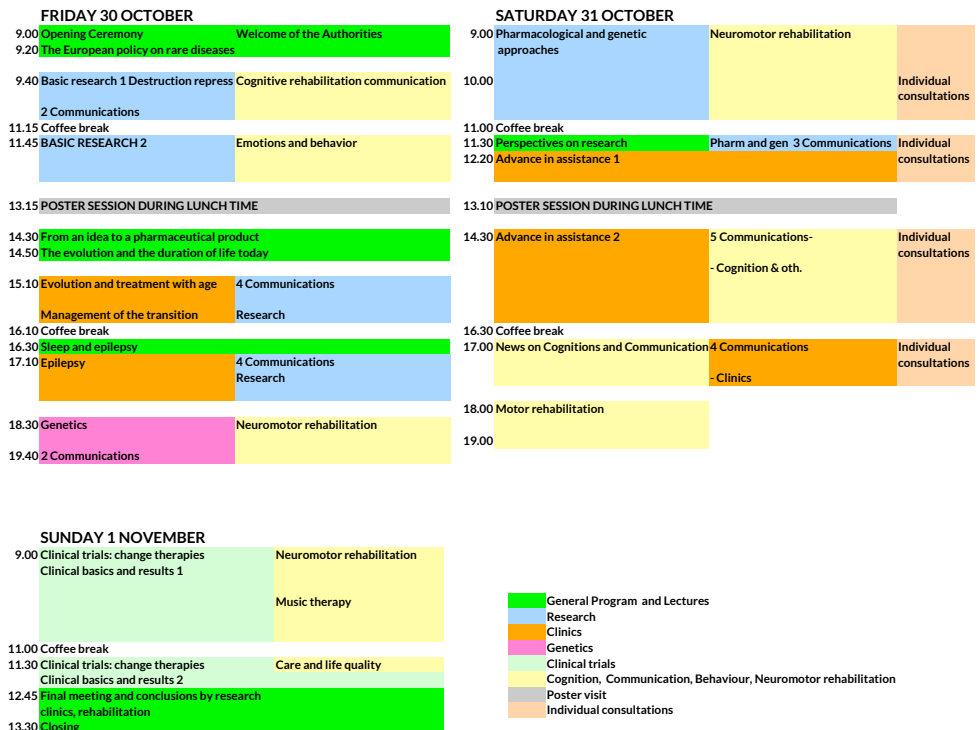
Room A and A - Aula B e A (See program for Therapists - Vedere programma terapisti).

Exhibition venues for aids, equipments and other medical devices - Spazi per esibizioni di apparecchi, ausili e altro.

Individual counseling for families about specific issues - Consulenze individuali per famiglie su specifici problemi.

Families are admitted at all the sessions - Le Famiglie sono ammesse a tutte le sessioni.

Workshop on the SUD in Rett Syndrome for clinicians of the Committee and representatives of Associations - Workshop sulla SUD nella Sindrome di Rett per clinici del Comitato e rappresentanti delle Associazioni.



PLENARY SESSION - SESSIONE PLENARIA

Room A - Aula A

Friday, October 30th - Venerdì 30 Ottobre

8.00-9.00

Registration - Registrazione

9.00

Opening Ceremony - Cerimonia di apertura

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

Welcome Remarks - Saluti delle Autorità

9.20

Lecture - Lezione magistrale

Chairman - Moderatore: Veneselli E (G Gaslini Institute, University of Genoa, Italy)

9.20 Taruscio D (ISS Rome, Italy)

Updates on the European policy about Rare Diseases - Update sulla politica europea per le Malattie Rare

9.40

Basic Research 1 - Ricerca di base 1

Chairmen - Moderatori: D'Esposito M (IGB Napoli, Italy) - Balas N (Stony Brook University, New York, USA)

9.40 Gabel HW (Washington University Medical School, USA)

Disruption of DNA-methylation-dependent long gene repression - Distruzione della repressione dipendente dalla metilazione del DNA di geni lunghi

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

Studying MeCP2 phosphorylation to obtain novel insights on MeCP2 functions and Rett syndrome - Lo studio della fosforilazione di MeCP2 come mezzo per ottenere nuove informazioni su MeCP2 e la sindrome di Rett

10.40-11.00 2 selected oral presentations - 2 presentazioni orali selezionate

MeCP2 is required for major satellite forward transcript recruitment at pericentric heterochromatin during neural differentiation

F. Della Ragione^{1,2}, S. Fioriniello¹, C. Zarrillo¹, M.R. Matarazzo¹, M. D'Esposito^{1,2}

¹Institute of Genetics and Biophysics "A Buzzati Traverso", Naples, Italy; ²Istituto di Ricovero e Cura a Carattere Scientifico Neuro-med, Pozzilli, Italy.

Novel human Rett syndrome in vitro models demonstrate differential impact of specific MeCP2 mutations on neuronal functions and the non-cell autonomous effects of glial cells

Bruria Ben Zeev¹, Amotz Ziv-Av², Shirtai Holtz², Simona Cazacu², Hae Kyung Lee³, Cunli Xiang³, Andreea Nissenkornm¹, Gila Kazimirsky² and Chaya Brodie^{2,3}

¹Pediatric Neurology Unit and Israeli Rett Clinic, Safra Pediatric Hospital, Sheba Med Ctr. Ramat-Gan, Israel; ²Everard and Mina Goodman Faculty of Life Sciences, Bar-Ilan University, Ramat-Gan, Israel; ³Davidson Laboratory of Cell Signaling and Tumor genesis, Department of Neurosurgery, Hermelin Brain Tumor Center, Henry Ford Hospital, Detroit, MI, USA

11.00 Discussion - Discussione

11.15

Coffee Break - Pausa caffè

11.45

Basic Research 2 - Ricerca di base 2

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

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

In vivo interrogation of MeCP2 using CRISPR-Cas9 - Interrogazione in vivo di MeCP2 usando CRISPR-Cas9

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

Glial cells, MECP2 and Rett Syndrome - Cellule gliali, MECP2 e Sindrome di Rett

12.35 Hupke P (University of Gottingen, Germany)

Wild-type microglia does not reverse pathology in mouse models of Rett syndrome - La microglia wild-type non reverte la patologia nei modelli murini della Sindrome di Rett

13.00 Discussion - Discussione

13.15

Lunch Break & Poster Viewing - Pausa pranzo & Visione dei poster

14.30

Lectures - Lezioni magistrali

Chairman - Moderatore: Curatolo P (University of Rome Tor Vergata, Italy)

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

From an idea to a pharmaceutical product - Da un'idea a un prodotto farmaceutico

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

Evolution over time and over the lifespan - Evoluzione nel tempo e della durata di vita

15.10

Evolution and Treatment from Childhood to Adulthood – Evoluzione e trattamento dall'infanzia all'età adulta

Chairmen - Moderatori: Budden S (Oregon Health & Science University, Oregon, USA) - Zappella M (University of Siena, Italy)

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

Protocols for Transitioning care from Childhood to Adulthood - Protocolli per la transizione delle cure dall'infanzia all'età adulta

15.30 Wilken B (Klinikum Kassel, Germany)

Management of Transition - Gestione della fase di transizione all'età adulta

15.50 Discussion - Discussione

16.10

Coffee Break – Pausa caffè

16.30

Lecture – Lezione magistrale

Chairman - Moderatore: Vigeveno F (Ospedale Pediatrico Bambino Gesù, Rome, Italy)

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

What we have learned about Epilepsy and Sleep - Cosa abbiamo imparato su Epilessia e Sonno

17.10

Focus on Epilepsy – Focus sull'Epilessia

Chairmen - Moderatori: Vigeveno F (Ospedale Pediatrico Bambino Gesù, Rome, Italy) - Pineda M (Hospital Sant Joan de Déu, Barcelona, Spain)

17.10 Nissenkorn A (Sourasky Medical Center, University of Tel Aviv, Israel)

Updates on Epilepsy - Nuove conoscenze sull'Epilessia

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

Epileptic and non-epileptic paroxysmal events - Disturbi parossistici epilettici e non-epilettici

17.50 Vignoli A (University of Milan, Italy)

Efficacy of new and alternative treatments - Efficacia dei trattamenti nuovi e alternativi

18.10 Discussion - Discussione

18.30

Genetics – Genetica

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

18.30 Russo S (Istituto Auxologico, Milan, Italy)

Exome sequencing of a cohort of Rett patients: new involved genes - Sequenziamento dell'esoma in una coorte di pazienti Rett: nuovi geni coinvolti

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

Improving genetic diagnosis using NGS: comparing exomes panels - Migliorare la diagnosi genetica usando NGS: confronto tra pannelli di esomi

19.10 2 selected oral presentations - 2 presentazioni orali selezionate

RNA sequencing analysis in iPSCs derived Rett neurons

Ilaria Meloni¹, Elisa Landucci¹, Laura Bianciardi¹, Sonia Amabile¹, Simone Furini², Flora Vaccarino^{3,4}, Valentina Imperatore¹, Caterina Lo Rizzo⁵, Maria Antonietta Mencarelli⁵, Elisa Frullanti¹, Francesca Ariani^{1,5}, Alessandra Renieri^{1,5}

¹Medical Genetics, University of Siena, Siena, Italy; ²Department of Medical Biotechnologies - University of Siena, Siena, Italy; ³Child Study Center, Yale University School of Medicine, New Haven, Connecticut, USA; ⁴Department of Neurobiology, Yale University School of Medicine, New Haven, Connecticut, USA; ⁵Genetica Medica, Azienda Ospedaliera Universitaria Senese, Siena, Italy

Discovery of new genes in Rett syndrome patients by WES

Vidal Falcó S¹, Lucariello, M², Vidal Ocabo, E², Brandi Tarrau NM³, Gerotina Mora E¹, Esteller Badosa M^{2,4}, Pineda Marfa M¹, Armstrong Morón J².

¹Fundación San Juan de Dios, Barcelona, ²Programa d'Epigenetica i Biologia del Cancer (PEBC), Institut d'Investigació Biomedica de Bellvitge (IDIBELL), ³Servicio de Genética Bioquímica&Rett, Hospital Universitario San Juan de Dios, Barcelona, ⁴Departament de Ciències Fisiològiques II, Facultat de Medicina, Universitat de Barcelona, Institució Catalana de Recerca i Estudis Avançats (ICREA), Barcelona, Catalonia, Spain.

19.30 Discussion - Discussione

Saturday, October 31st - Sabato 31 ottobre

8.00-9.00

Registration – Registrazione

9.00

Pharmacological and Genetic Approaches 1 – Approcci farmacologici e genetici 1

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

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

Genetic and Pharmacological Reactivation of

the Mammalian Inactive X Chromosome: Implications for Treatment - Riattivazione genetica e farmacologica del cromosoma X mammaliano inattivo: implicazioni per il trattamento

9.25 Cobb S (University of Glasgow, UK)

Gene Therapy - Terapia genica

9.50 Discussion - Discussione

10.00 Ciani E (University of Bologna, Italy)

Therapies for CDKL5 disorder - Terapie per il disordine CDKL5

10.25 Giustetto M (University of Turin, Italy)
Synaptic and behavioural phenotypes in MECP2 and CDKL5 mouse models: insights into pharmacological therapies - Alterazioni dei circuiti nervosi e del comportamento nei modelli murini mutanti per MECP2 e CDKL5: risultati di terapie farmacologiche precliniche

10.50 Discussion - Discussione

11.00

Coffee Break - Pausa Caffé

11.30

Perspectives on research - Prospettive di ricerca

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

11.30 De Montleau B (Eurordis)

Supporting research in Europe - Supportare la ricerca in Europa

11.50 Mari F (University of Siena, Italy)

Updates on Rett Networked Database - Update sul Rett Networked Database

12.10 Discussion - Discussione

12.20

Advance in Assistance and Complex Issues: Care and Prevention 1 - Avanzamento dell'assistenza e problematiche complesse: Care e Prevenzione 1

Chairman - Moderatore: Canevini M.P (University of Milan, Italy)

12.20 Budden S (Oregon Health & Science University, Oregon, USA)

Recognition and Management of Autonomic Disturbance - Riconoscimento e gestione dei Disturbi autonomici

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

Improvements in Emergency Assistance: the Clinical Passport - Nuovi avanzamenti dell'assistenza in Emergenza: il Passaporto Clinico

13.00 Discussion - Discussione

13.10

Lunch Break & Poster Viewing - Pausa pranzo & Visione dei poster | General Assembly RSE - Assemblea Generale RSE

14.30

Advance in Assistance and Complex Issues: Care and Prevention 2 - Avanzamento dell'assistenza e problematiche complesse: Care e Prevenzione 2

Chairmen - Moderatori: Wilken B (Klinikum Kassel, Germany), Papoff P (La Sapienza University, Rome)

14.30 Kendrick A (University Hospitals, Bristol, UK)

Breathing problems and their diagnosis - I disturbi del respiro e la loro diagnosi

14.50 Chiarini Testa MB (Ospedale Pediatrico Bambino Gesù, Rome, Italy)

Breathing problems and their treatment (management)- I disturbi del respiro e il loro trattamento

15.10 Discussion - Discussione

15.20 Canevini MP (University of Milan, Italy)

The management of epilepsy in complex disabilities - La gestione dell'epilessia nelle disabilità complesse

15.40 Rimini A (G Gaslini Institute, Genoa, Italy), Mancardi MM (G Gaslini Institute, Genoa, Italy)

Preventive Cardiology (cardiological prevention) and drug interactions - Cardiologia preventiva e interazioni tra farmaci

16.00 Cefalo G (University of Milan, Italy)

The management of nutrition related to age and disability - La gestione della nutrizione correlata all'età e alla disabilità

16.20 Discussion - Discussione

16.30

Coffee Break - Pausa caffè

17.00

New Insights in Cognition and Communication - Nuove idee sui Processi cognitivi e sulla Comunicazione

Chairmen - Moderatori: Fabio RA (University of Messina, Italy) - Leuzzi V (La Sapienza University of Rome, Italy)

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

Communication and cognitive empowerment - Comunicazione e potenziamento cognitivo

17.20 Fabio RA (University of Messina, Italy)

On the bridge between neurological and psychological factors: TdCS and cognitive empowerment - A ponte tra fattori neurologici e psicologici: TdCS e potenziamento cognitivo

17.40 Discussion - Discussione

18.00

Movement Disorders: Evaluation and Rehabilitation - Disturbi del Movimento: Valutazione e Riabilitazione

Chairmen - Moderatori: Budden S (Oregon Health & Science University, Oregon, USA) - Rodocanachi M (Fondazione Don Gnocchi, Milan, Italy)

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

Motor development: new frontiers for walking activity evaluation - Sviluppo Motorio: nuove frontiere per la valutazione del cammino

18.20 Lotan M (Ariel University, Israel)

Rehabilitation: advances and new trials - Riabilitazione: avanzamenti e nuovi trial

18.40 - 19.00 Discussion - Discussione

9.00

Focus on Clinical Trials: From Scientific Background to Results; How to Change the Therapies 1 – Focus sui Trial Clinici: dalle basi scientifiche ai risultati; come modificare le terapie 1

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

9.00 Kaufmann W (Harvard Med. School, USA)

The lessons from IGF-1 trials - Lezioni dai Trial con IGF-1

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

NNZ-2566: a novel. experimental treatment - NNZ-2566: un trattamento sperimentale nuovo

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

Pharmacological treatments with Copaxone and Statins - Trattamenti farmacologici con Copaxone e Statine

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

Copaxone from cells to RTT patients - hopes and unexpected adverse events - Il copaxone dagli studi cellulari alle pazienti Rett: speranze e eventi avversi inattesi

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

Desipramine and cardio-respiratory variability: results of a clinical trial in breathing disorders - Desipramina e variabilità cardio-respiratoria: risultati di un trial clinico sui disturbi del respiro

10.40 Discussion - Discussione

11.00-11.30

Coffee Break - Pausa caffè

11.30

Focus on Clinical Trials: From Scientific Background to Results; How to Change the Therapies 2 – Focus sui Trial Clinici: dalle basi scientifiche ai risultati; come modificare le terapie 2

11.30 Rothenbuhler A. (Hôpital Bicêtre, Paris, France)

Bone Health in Rett Syndrome - La salute delle ossa nella Sindrome di Rett

11.50 Hayek J. (Policlinico Le Scotte, Siena, Italy)

EPI-743 trial results - Risultati del trial con EPI-743

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

Rethinking the future of clinical trials in Rett Syndrome - Ripensare il futuro dei trial clinici nella Sindrome di Rett

12.30 Discussion - Discussione

12.45

Final Meeting and Experts Conclusions – Meeting finale e conclusioni degli esperti

12.45 Research experts final considerations - Considerazioni conclusive degli esperti in ricerca

13.00 Clinic experts final considerations - Considerazioni conclusive degli esperti in clinica

13.15 Rehabilitation experts final considerations - Considerazioni conclusive degli esperti in riabilitazione

13.30

Closing remarks - Chiusura dei lavori

ECM
856-136115

Sono previsti n° 15 ECM crediti ECM per la figura professionale di:

Medico Chirurgo (tutte le discipline), Farmacista, Biologo, Infermiere, infermiere pediatrico.

La presenza dei partecipanti sarà verificata tramite firma di entrata e uscita e compilazione della scheda di valutazione dell'evento. Per avere diritto ai crediti è necessario frequentare il 100% delle ore di formazione. La verifica dell'apprendimento avverrà tramite questionario con domande a risposta multipla.

Obiettivi formativi di sistema: N°2 Linee guida - protocolli - procedure.

CME accreditation
856-136115

15 Italian CME credits will be available for medical doctors (all disciplines), pharmacist, biologists, nurses, pediatric nurses.

The participant's CME credit will be dependent upon completion and return of a content specific quiz to test the knowledge after the CME activity, and the satisfaction form.

PROVIDER

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16148 Genova
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www.cisef.org



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PARALLEL SESSION - SESSIONE PARALLELA

Room B – Aula B

Friday, October 30th - Venerdì 30 Ottobre

9.45

Cognitive Empowerment and Rehabilitation of Communication – Potenziamento Cognitivo e Riabilitazione della Comunicazione

Chairman - Moderatore: Budden S. (Oregon Health & Science University, Oregon, USA)

9.45 Marschik, P. (University of Gratz, Austria)

Recognizing atypical developmental pathways: implications for speech, language and communication intervention - Riconoscere i pathways di sviluppo atipico: implicazioni per il linguaggio e la comunicazione

10.10 Fabio R.A. (University of Messina, Italy)

Empowerment of attention, memory and communication - Potenziamento di attenzione, memoria e comunicazione

10.35 Giannatiempo S. (TICE, Piacenza, Italy)

New Learning software, Eyetracker, the tablet as an instrument to play and communicate - Nuovo software su Apprendimento, Eyetracker, uso del tablet per giocare e comunicare

11.00 Discussion - Discussione

11.15

Coffee Break – Pausa caffè

11.45

Emotions and Behaviour – Le Emozioni e il Comportamento

Chairman - Moderatore: Vignoli A. (University of Milan, Italy)

11.45 Budden S. (Oregon Health & Science University, Oregon, USA)

Emotional and Behavioral Management - Trattamento emozionale e del comportamento

12.15 Fabio RA (University of Messina, Italy)

How to manage behavioural problems - Come gestire i problemi di comportamento

12.45 - 13.15 Discussion - Discussione

13.15

Pausa pranzo & Visione dei poster

15.10-16.10

Basic Research

Chairman - Moderatore: Giustetto M (University of Turin, Italy)

4 selected oral presentations - 4 presentazioni orali selezionate

Phosphorylation of huntingtin at Ser421 modulates the phenotype of the Mecp2-deficient mice

Yann Ehinger^{1,2}, Lydia Saidj^{1,2}, Valérie Matagne^{1,2}, Julie Bruyère^{3,4}, Frederic Saudou^{3,5}, Laurent Villard^{1,2}, JeanChristophe Roux^{1,2}

¹Inserm, UMR_S 910, Faculté de Médecine de La Timone, Marseille, France; ²Aix Marseille Université, GMGF, Marseille, France; ³Inserm, U836, F-38000 Grenoble, France; ⁴Université Grenoble Alpes, Grenoble Institut des Neurosciences, GIN, Grenoble, France; ⁵CHU de Grenoble, Grenoble, France.

A mouse model of neuronal-specific MeCP2 overexpression

Martha V. Koerner, Jim Selfridge and Adrian P. Bird

Wellcome Trust Centre for Cell Biology, University of Edinburgh, UK

Gastrointestinal dysmotility in a mouse model of Rett syndrome

Sarah Schock¹, George Wahba², William Staines², David Grynspan³

¹Children's Hospital of Eastern Ontario, Research Institute, Ottawa, Ontario, Canada; ²University of Ottawa, Department of Cellular and Molecular Medicine, Ottawa, Ontario, Canada; ³Children's Hospital of Eastern Ontario, Pathology, Ottawa, Ontario, Canada

Full-length recombinant production of human CDKL5 protein in Antarctic marine bacterium Pseudoalteromonas haloplanktis

Valeria Citarella, Filomena Sannino, Ermenegilda Parrilli, and Maria Luisa Tutino

Dipartimento di Scienze Chimiche, Università di Napoli Federico II- Complesso Universitario M. S. Angelo, via Cintia, 80126 Napoli, Italia

17.10-18.30

Pharmacological and genetic approaches

Chairmen - Moderatori: Laviola G (ISS, Rome, Italy) - Renieri A (University of Siena, Italy)

4 selected oral presentations - 4 presentazioni orali selezionate

The cell-permeable JNK inhibitor peptide (D-JN-KI1) prevents motor defects and preserves dendritic spines in Mecp2 and CDKL5 mice models
Sara Cimini¹, Eleonora Calcagno⁴, Anna Maria Castaldo¹, Grazia Della Sala², Elena Putignano³, Tommaso Pizzorusso³, Maurizio Giustetto⁴, Tiziana Borsello¹

¹IRCCS Istituto di Ricerche Neurologiche Mario Negri, Milano; ²Department of NEUROFARBA University of Florence, Florence, Italy; ³Institute of Neuroscience, CNR, Pisa, Italy; ⁴University of Turin, Department of Neuroscience and National Institute of Neuroscience, Turin, Italy.

Pharmacological treatment with Mirtazapine rescues cortical atrophy and respiratory deficits in

MeCP2 null mice

Tongiorgi Enrico; Bittolo Tamara, Raminelli Carlo Antonio, Deiana Chiara, Baj Gabriele, Vaghi Valentina, Ferrazzo Sara, Bernareggi Annalisa

Department of Life Sciences, University of Trieste, Italy

MECP2 gene therapy for Rett syndrome: adding regulation

Ralph D. Hector^{1,2}, Andrew McCracken^{1,2}, Mark E.S. Bailey², Stuart R. Cobb¹

¹Institute of Neuroscience and Psychology, and ²School of Life Sciences, College of Medical, Veterinary and Life Sciences, University of Glasgow

Evaluating the effect of new readthrough drugs using a novel in vitro Rett Syndrome system, human neural cells carrying MeCP2 nonsense mutations

Bruria Ben-Zeev¹, Andreea Nissenkorn¹, Timor Baa-sov², Meytal Shohat³, Rinat Sack³, Shmuel Tuvia³ and Chaya Brodie^{4,5}

¹Pediatric Neurology Unit and Israeli Rett Clinic, Safra Pediatric Hospital, Sheba Med Ctr. Ramat-Gan, Israel; ²Edith and Joseph Fischer Enzyme Inhibitors Laboratory, Schulich Faculty of Chemistry, Technion IIT, Haifa, Israel; ³Eloxx Pharmaceuticals, Nes Ziona,

Israel; ⁴Davidson Laboratory of Cell Signaling and Tumorigenesis, Department of Neurosurgery, Hermelin Brain Tumor Center, Henry Ford Hospital, Detroit, MI, USA; ⁵Everard and Mina Goodman Faculty of Life Sciences, Bar-Ilan University, Ramat-Gan, Israel

18.30

Neuromotor Rehabilitation - Riabilitazione Neuromotoria

Chairman - Moderatore: Cini C. (Ospedale Pediatrico Bambino Gesù, Rome, Italy)

18.30 Cosentino A. (Don Calabria Institute, Verona, Italy)

Design for all: projects for sensorial and sensorimotor stimulation - Progetti per la stimolazione sensoriale e sensomotoria

18.50 Coluccini M. (University of Pisa, Italy)

Advanced walking devices to train posture and locomotion in children with neuromotor disabilities - Ausili di nuova generazione per allenare la postura ed il cammino in bambini con disabilità neuromotoria

19.10- 19.30 Discussion - Discussione

Saturday, October 31st - Sabato 31 Ottobre

9.00

Neuromotor Rehabilitation - Riabilitazione Neuromotoria

Chairman - Moderatore: Castelli E. (Ospedale Pediatrico Bambino Gesù, Rome, Italy)

9.00 Rodocanachi M. (Fondazione Don Gnocchi, Milan, Italy)

Proposal for a new grid to observe gross motor modifiability - Proposta di una griglia di osservazione per la modificabilità motoria

9.20 Lotan M. (Ariel University, Israel)

Losing and regaining walking ability - Perdere e recuperare la capacità di deambulare

9.40- 10.00 Discussion - Discussione

10.00-10.20 Rodocanachi M. (Fondazione Don Gnocchi, Milan, Italy)

Rehabilitation of gait initiation: from gait analysis to clinical practice - Riabilitazione dell'inizio della deambulazione: dall'analisi del cammino alla pratica clinica

10.20-10.40 Lotan M. (Ariel University, Israel)

Enhancing walking ability in the adult patient through Applied Behaviour Analysis (ABA) - Migliorare capacità di camminare in età adulta attraverso l'analisi applicata del comportamento (ABA)

10.40- 11.00 Discussion - Discussione

11.00

Coffee Break - Pausa Caffè

11.30-12.20

Pharmacological and Genetic Approaches 2 - Approcci farmacologici e genetici 2

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

3 selected oral presentations - 3 presentazioni orali selezionate

Dendritic Spine Instability in a Mouse Model of CDKL5 Disorder is rescued by IGF-1

*G.Della Sala^{*1}, E. Putignano^{*2}, G. Chelini¹, R. Melani¹, E. Calcagno³, G.M. Ratto⁴, E. Amendola⁵, C. T. Gross⁵, M. Giustetto^{E3}, T. Pizzorusso^{E1,2}*

¹Department of NEUROFARBA University of Florence, Area San Salvi - Pad. 26, 50135 Florence, Italy; ²Institute of Neuroscience, CNR, Via Moruzzi 1, 56124 Pisa, Italy; ³University of Turin, Department of Neuroscience and National Institute of Neuroscience, Corso M. D'Azeglio 52, 10126 Turin, Italy; ⁴NEST, Institute of Nanoscience CNR and Scuola Normale Superiore, Pisa, Italy; ⁵Mouse Biology Unit, European Molecular Biology Laboratory (EMBL), via Ramarini 32, 00015 Monterotondo, Italy

In vivo treatment of Mecp2^{-y} mice with the radical scavenger Trolox

Oliwia A. Janc, Marc Hüser, Karolina Can, Belinda Kempkes, and Michael Müller

Zentrum Physiologie und Pathophysiologie, Institut für Neuro- und Sinnesphysiologie, Universitätsmedizin Göttingen, Göttingen, Germany

Targeting brain Rho GTPases in Rett Syndrome: Preclinical evidence of therapeutic effects

De Filippis, Bianca^a, Ricceri, Laura^b, Laviola, Giovanni^a

^aBehavioural Neuroscience Section, Dept. Cell Biology & Neuroscience, Istituto Superiore di Sanità, Roma, Italy; ^bNeurotoxicology and Neuroendocrinology Imaging Section, Dept. Cell Biology & Neuroscience, Istituto Superiore di Sanità, Roma, Italy

12.10-12.20 Discussion - Discussione

14.30-16.30

Cognition and Communication

Chairmen - Moderatori: Fabio RA (University of Messina, Italy)
- Giannatiempo S (TICE, Piacenza, Italy)

5 selected oral presentations - 5 presentazioni orali selezionate

Tools for Assessment of Communication in Rett Syndrome

Helena Wandin^a, Theresa Bartolotta^b, Sally-Ann Garrett^c, Anna Urbanowicz^d, Gillian Townend^e

^aSwedish National Rett Center, Frösön, Sweden; ^bSchool of Health Sciences, Stockton University, Galloway, NJ, USA; ^cIsle Talk, Kiltorglin, County Kerry, Republic of Ireland; ^dUniversity of Western Australia & Edith Cowan University, Perth, WA, Australia; ^eRett Expertise Centre - GKC, Maastricht University Medical Centre, Maastricht, The Netherlands

Presenting authors: Helena Wandin, Theresa Bartolotta, Gillian Townend

Typical or Atypical? The Intermittent Character of Early Vocalizations in the Preserved Speech Variant

Florian B Pokorny^{1,2,3}, Katrin D Bartl-Pokorny¹, Dajie Zhang¹, Ralf Vollmann^{3,4}, Christa Einspieler¹, Peter B Marschik^{1,3,5}

¹Research Unit iDN - interdisciplinary Developmental Neuroscience, Institute of Physiology, Medical University of Graz, Austria; ²Machine Intelligence & Signal Processing group, Technische Universität München, Germany; ³Brain, Ears & Eyes - Pattern Recognition Initiative, BioTechMed-Graz, Austria; ⁴Department of Linguistics, University of Graz; ⁵Center of Neurodevelopmental Disorders, Karolinska Institutet, Stockholm, Sweden

Activities for interaction; examples of group activities with special focus on communication with PODD (Pragmatic Organisation Dynamic Displays)

Helena Wandin, Märith Bergström-Isacson

Swedish National Rett Center, Frösön, Sweden

Eye gaze technology as a form of augmentative and alternative communication support for individuals with Rett syndrome: experiences of families in the Netherlands

Gillian S. Townend^a, Eric Smeets^a, Raymond van de Berg^{a,b,c}, Mariëlle van den Berg^d, Dick van Waardenburg^a & Leopold M.G. Curfs^a

^aRett Expertise Centre - GKC, Maastricht University Medical Centre, Maastricht, The Netherlands; ^bDivision of Balance Disorders, Department of Otorhinolaryngology and Head and Neck Surgery, Faculty of Health Medicine and Life Sciences, School for Mental Health and Neuroscience, Maastricht University Medical Center, Maastricht, The Netherlands; ^cFaculty of Physics, Tomsk State University, Russian Federation; ^dNederlandse Rett Syndroom Vereniging, Dutch Rett Syndrome Parent Association, Utrecht, The Netherlands

Using Eye-tracking Technology in Assessing Receptive Language Skills among Girls with Rett Syndrome

Ahonniska-Assa, J., Silberg T., Pollak, O., Saraf, E., Wine, J., Nissenkorn, A., & Ben-Zeev, B.

Neuropsychology Unit, Pediatric Rehabilitation Department, Edmond and Lily Safra Children's Hospital, The Chaim Sheba Medical Center; Pediatric Neurology Unit, Edmond and Lily Safra Children's Hospital, The Chaim Sheba Medical Center; Pediatric Neurology Unit, Edmond and Lily Safra Children's Hospital, The Chaim Sheba Medical Center

16.30-17.00

Coffee Break - Pausa Caffè

17.00-18.00

Clinics

Chairmen - Moderatori: Vignoli A (University of Milan, Italy) - Specchio N (Ospedale Bambino Gesù, Roma, Italy)

4 selected oral presentations - 4 presentazioni orali selezionate

Can girls with Rett syndrome (re)learn gross motor skills after regression?

Anne-Marie Bisgaard, Bitten Schönewolf-Greulich, Birgit Syhler, Michelle Stahlhut

Centre for Rett syndrome, Department of Clinical Genetics - Kennedy Centre, Rigshospitalet, Copenhagen, Denmark

Study of Rett syndrome epidemiology and database in Japan

Masayuki Itoh¹, Shin Nabatame², Hisateru Tachimori¹, Toyojiro Matsuishi^{3,4}

Consortium for Japanese Rett syndrome survey, ¹National Center of Neurology and Psychiatry, ²Osaka University, ³Kurume University, ⁴St. Mary's Hospital, Japan.

Efficacy and tolerability of antiepileptic drugs in 104 girls with Rett syndrome

Savini MN, Vignoli A, Nowbut MS, Peron A, Turner K, La Briola F, Canevini MP

Epilepsy Center, San Paolo Hospital, Health Sciences Department, University of Milan, Italy

Cytokine Dysregulation in MECP2- and CDKL5-Rett Syndrome

Joussef Hayek¹, Silvia Leoncini^{1,2}, Cinzia Signorini², Gloria Zollo^{1,2}, Alessio Cortelazzo^{1,3}, Thierry Durand⁴, Jean-Marie Galano⁴, Roberto Guerranti³, Lucia Ciccoli², Claudio De Felice⁵

¹Child Neuropsychiatry Unit, University Hospital, Azienda Ospedaliera Universitaria Senese (AOUS), Policlinico "S.M. alle Scotte", Siena, Italy ²Department of Molecular and Developmental Medicine, University of Siena, Siena, Italy ³Department of Medical Technologies, University of Siena, 53100 Siena, Italy ⁴Institut des Biomolécules Max Mousseron (IBMM), UMR 5247, CNRS/UM/ENSCM, Montpellier, France ⁵ Neonatal Intensive Care Unit, University Hospital, AOUS, Policlinico "S. M. alle Scotte", Siena, Italy

Sunday, November 1st - Domenica 1 Novembre

9.00

Neuromotor Rehabilitation – Riabilitazione Neuromotoria

Chairman - Moderatore: Rodocanachi M (Fondazione Don Gnocchi, Milan, Italy)

9.00 Lotan M. (Ariel University, Israel)

Conventional physical therapy for scoliosis - Terapia fisica convenzionale per la scoliosi

9.25 Lotan M. (Ariel University, Israel)

Hand mannerism and scoliosis is there a link? - Manierismo della mano e scoliosi, c'è un legame?

9.50 - 10.00 Discussion - Discussione

10.00

Music Therapy – Musicoterapia

Chairman - Moderatore: Pintaudi M (IGG, University of Genoa, Italy)

10.00 Cochavit E. (Ariel University, Israel)

How helping girls with music - Come aiutare le bambine con la musica

10.25 Cochavit E. (Ariel University, Israel)

Music and Augmentative Communication - Musica e Comunicazione Aumentativa

10.50 - 11.00 Discussion - Discussione

11.00-11.30

Coffee Break – Pausa caffè

11.30

Care and Quality of Life – “Care” e Qualità di Vita

Chairman - Moderatore: Veneselli E (G Gaslini Institute, University of Genoa, Italy)

11.30 Biondi D. (Don Calabria Institute, Rome, Italy), Rodocanachi M. (Fondazione Don Gnocchi, Milan, Italy)

Psychological care and quality of life - “Care” psicologica e qualità di vita

11.50 - 12.10 Discussion – Discussione

PROGRAM FOR THERAPISTS, EDUCATORS AND TEACHERS
PROGRAMMA PER TERAPISTI, EDUCATORI E INSEGNANTI



In Room A - in Aula A



In Room B - in Aula B

Friday, October 30th - Venerdì 30 Ottobre

8.00-9.00

Registration – Registrazione

9.00

Opening Ceremony – Cerimonia di apertura

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

Welcome Remarks – Saluti delle Autorità

9.20

Lecture – Lezione magistrale

Chairman - Moderatore: Veneselli E (G Gaslini Institute, University of Genoa, Italy)

9.20 Taruscio D (ISS Rome, Italy)

Updates on the European policy about Rare Diseases - Update sulla politica europea per le Malattie Rare

9.45

Cognitive Empowerment and Rehabilitation of Communication – Potenziamento Cognitivo e Riabilitazione della Comunicazione

Chairman - Moderatore: Budden S. (Oregon Health & Science University, Oregon, USA)

9.45 Marschik, P. (University of Gratz, Austria)

Recognizing atypical developmental pathways:

implications for speech, language and communication intervention - Riconoscere i pathways di sviluppo atipico: implicazioni per il linguaggio e la comunicazione

10.10 Fabio R.A. (University of Messina, Italy)

Empowerment of attention, memory and communication - Potenziamento di attenzione, memoria e comunicazione

10.35 Giannatiempo S. (TICE, Piacenza, Italy)

New Learning software, Eyetracker, the tablet as an instrument to play and communicate - Nuovo software su Apprendimento, Eyetracker, uso del tablet per giocare e comunicare

11.00 Discussion - Discussione

11.15

Coffee Break – Pausa caffè

11.45

Emotions and Behaviour – Le Emozioni e il Comportamento

Chairman - Moderatore: Vignoli A (University of Milan, Italy)

11.45 Budden S. (Oregon Health & Science University, Oregon, USA)

Emotional and Behavioral Management - Trattamento emozionale e del comportamento

12.15 Fabio RA (University of Messina, Italy)
How to manage behavioural problems - Come gestire i problemi di comportamento

12.45 - 13.15 Discussion - Discussione

13.15

Lunch Break & Poster Viewing – Pausa pranzo & Visione dei poster

14.30

Lectures – Lezioni magistrali

Chairman - Moderatore: Curatolo P (University of Rome Tor Vergata, Italy)

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

From an idea to a pharmaceutical product - Da un'idea a un prodotto farmaceutico

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

Evolution over time and over the lifespan - Evoluzione nel tempo e della durata di vita

15.10

Evolution and Treatment from Childhood to Adulthood – Evoluzione e trattamento dall'infanzia all'età adulta

Chairmen - Moderatori: Budden S (Oregon Health & Science University, Oregon, USA) - Zappella M (University of Siena, Italy)

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

Protocols for Transitioning care from Childhood to Adulthood - Protocolli per la transizione delle cure dall'infanzia all'età adulta

15.30 Wilken B (Klinikum Kassel, Germany)

Management of Transition - Gestione della fase di transizione all'età adulta

15.50 Discussion - Discussione

16.10

Coffee Break – Pausa caffè

16.30

Lecture – Lezione magistrale

Chairman - Moderatore: Vigeveno F (Ospedale Pediatrico Bambino Gesù, Rome, Italy)

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

What we have learned about Epilepsy and Sleep - Cosa abbiamo imparato su Epilessia e Sonno

17.10

Focus on Epilepsy – Focus sull'Epilessia

Chairmen - Moderatori: Vigeveno F (Ospedale Pediatrico Bambino Gesù, Rome, Italy) - Pineda M (Hospital Sant Joan de Déu, Barcelona, Spain)

17.10 Nissenkorn A (Sourasky Medical Center, University of Tel Aviv, Israel)

Updates on Epilepsy - Nuove conoscenze sull'Epilessia

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

Epileptic and non-epileptic paroxysmal events - Disturbi parossistici epilettici e non-epilettici

17.50 Vignoli A (University of Milan, Italy)

Efficacy of new and alternative treatments - Efficacia dei trattamenti nuovi e alternativi

18.10 Discussion - Discussione

18.30

Neuromotor Rehabilitation – Riabilitazione Neuromotoria

Chairman - Moderatore: Cini C (Ospedale Pediatrico Bambino Gesù, Rome, Italy)

18.30 Cosentino A. (Don Calabria Institute, Verona, Italy)

Design for all: projects for sensorial and sensorimotor stimulation - Progetti per la stimolazione sensoriale e sensomotoria

18.50 Coluccini M. (University of Pisa, Italy)

Advanced walking devices to train posture and locomotion in children with neuromotor disabilities - Ausili di nuova generazione per allenare la postura ed il cammino in bambini con disabilità neuromotoria

19.10 - 19.30 Discussion – Discussione

Saturday, October 31st - Sabato 31 Ottobre

8.00-9.00

Registration – Registrazione

9.00

Neuromotor Rehabilitation – Riabilitazione Neuromotoria

Chairman - Moderatore: Castelli E (Ospedale Pediatrico Bambino Gesù, Rome, Italy)

9.00 Rodocanachi M. (Fondazione Don Gnocchi, Milan, Italy)

Proposal for a new grid to observe gross motor modifiability - Proposta di una griglia di osserva-

zione per la modificabilità motoria

9.20 Lotan M. (Ariel University, Israel)

Losing and regaining walking ability - Perdere e recuperare la capacità di deambulare

9.40- 10.00 Discussion – Discussione

10.00-10.20 Rodocanachi M. (Fondazione Don Gnocchi, Milan, Italy)

Rehabilitation of gait initiation: from gait analysis to clinical practice - Riabilitazione dell'inizio della deambulazione: dall'analisi del cammino alla pratica clinica

10.20-10.40 Lotan M. (Ariel University, Israel)
Enhancing walking ability in the adult patient through Applied Behaviour Analysis (ABA) - Migliorare capacità di camminare in età adulta attraverso l'analisi applicata del comportamento (ABA)

10.40- 11.00 Discussion – Discussione

11.00

Coffee Break – Pausa Caffè

11.30

Perspectives on research – Prospettive di ricerca

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

11.30 De Montleau B (Eurordis)

Supporting research in Europe - Supportare la ricerca in Europa

11.50 Mari F (University of Siena, Italy)

Updates on Rett Networked Database - Update sul Rett Networked Database

12.10 Discussion - Discussione

12.20

Advance in Assistance and Complex Issues: Care and Prevention 1 – Avanzamento dell'assistenza e problematiche complesse: Care e Prevenzione 1

Chairman - Moderatore: Canevini MP (University of Milan, Italy)

12.20 Budden S (Oregon Health & Science University, Oregon, USA)

Recognition and Management of Autonomic Disturbance - Riconoscimento e gestione dei Disturbi autonomici

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

Improvements in Emergency Assistance: the Clinical Passport - Nuovi avanzamenti dell'assistenza in Emergenza: il Passaporto Clinico

13.00 Discussion - Discussione

13.10

Lunch Break & Poster Viewing – Pausa pranzo & Visione dei poster | General Assembly RSE - Assemblea Generale RSE

14.30

Advance in Assistance and Complex Issues: Care and Prevention 2 - Avanzamento dell'assistenza e problematiche complesse: Care e Prevenzione 2

Chairmen - Moderatori: Wilken B (Klinikum Kassel, Germany), Papoff P (La Sapienza University, Rome)

14.30 Kendrick A (University Hospitals, Bristol, UK)

Breathing problems and their diagnosis - I disturbi del respiro e la loro diagnosi

14.50 Chiarini Testa MB (Ospedale Pediatrico Bambino Gesù, Rome, Italy)

Breathing problems and their treatment (management)- I disturbi del respiro e il loro trattamento

15.10 Discussion - Discussione

15.20 Canevini MP (University of Milan, Italy)

The management of epilepsy in complex disabilities - La gestione dell'epilessia nelle disabilità complesse

15.40 Rimini A (G Gaslini Institute, Genoa, Italy),

Mancardi MM (G Gaslini Institute, Genoa, Italy)
Preventive Cardiology (cardiological prevention) and drug interactions - Cardiologia preventiva e interazioni tra farmaci

16.00 Cefalo G (University of Milan, Italy)

The management of nutrition related to age and disability - La gestione della nutrizione correlata all'età e alla disabilità

16.20 Discussion - Discussione

16.30

Coffee Break – Pausa caffè

17.00

New Insights in Cognition and Communication – Nuove idee sui Processi cognitivi e sulla Comunicazione

Chairmen - Moderatori: Fabio RA (University of Messina, Italy) - Leuzzi V (La Sapienza University of Rome, Italy)

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

Communication and cognitive empowerment - Comunicazione e potenziamento cognitivo

17.20 Fabio RA (University of Messina, Italy)

On the bridge between neurological and psychological factors: TdCS and cognitive empowerment - A ponte tra fattori neurologici e psicologici: TdCS e potenziamento cognitivo

17.40 Discussion - Discussione

18.00

Movement Disorders: Evaluation and Rehabilitation – Disturbi del Movimento: Valutazione e Riabilitazione

Chairmen - Moderatori: Budden S (Oregon Health & Science University, Oregon, USA) - Rodocanachi M (Fondazione Don Gnocchi, Milan, Italy)

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

Motor development: new frontiers for walking activity evaluation - Sviluppo Motorio: nuove frontiere per la valutazione del cammino

18.20 Lotan M (Ariel University, Israel)

Rehabilitation: advances and new trials - Riabilitazione: avanzamenti e nuovi trial

18.40 - 19.00 Discussion - Discussione

9.00

Neuromotor Rehabilitation - Riabilitazione Neuromotoria

Chairman - Moderatore: Rodocanachi M (Fondazione Don Gnocchi, Milan, Italy)

9.00 Lotan M. (Ariel University, Israel)

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Chairman - Moderatore: Pintaudi M (IGG, University of Genoa, Italy)

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Coffee Break - Pausa caffè

11.30

Care and Quality of Life - "Care" e Qualità di Vita

Chairman - Moderatore: Veneselli E (G Gaslini Institute, University of Genoa, Italy)

11.30 Biondi D. (Don Calabria Institute, Rome, Italy), Rodocanachi M. (Fondazione Don Gnocchi, Milan, Italy)

Psychological care and quality of life - "Care" psicologica e qualità di vita

11.50 - 12.10 Discussion - Discussione

12.10

Focus on Clinical Trials: From Scientific Background to Results; How to Change the Therapies 2 - Focus sui Trial Clinici: dalle basi scientifiche ai risultati; come modificare le terapie 2

12.10 Nguyen G (Hôpital Avicenne - Hôpitaux

Universitaires Paris-Seine-Saint-Denis, France) Rethinking the future of clinical trials in Rett Syndrome - Ripensare il futuro dei trial clinici nella Sindrome di Rett

12.30 Discussion - Discussione

12.45

Final Meeting and Experts Conclusions - Meeting finale e conclusioni degli esperti

12.45 Research experts final considerations - Considerazioni conclusive degli esperti in ricerca

13.00 Clinic experts final considerations - Considerazioni conclusive degli esperti in clinica

13.15 Rehabilitation experts final considerations - Considerazioni conclusive degli esperti in riabilitazione

13.30

Closing remarks - Chiusura dei lavori

ECM

856-136118

Sono previsti n° 15 ECM crediti ECM per la figura professionale di:

Fisioterapista, Terapista della Neuro e Psicomotricità dell'età evolutiva, Logopedista, Psicologo, Educatore professionale

La presenza dei partecipanti sarà verificata tramite firma di entrata e uscita e compilazione della scheda di valutazione dell'evento. Per avere diritto ai crediti è necessario frequentare il 100% delle ore di formazione. La verifica dell'apprendimento avverrà tramite questionario con domande a risposta multipla.

Obiettivi formativi di sistema: N°2 Linee guida - protocolli - procedure

Il Programma integra le tematiche cliniche e riabilitative sviluppate in plenaria con una serie di sessioni su: potenziamento di comunicazione, attenzione, memoria, anche con nuovi software; focus sulla gestione dei problemi emozionali e di comportamento nelle varie età; le ultimissime novità tecnologiche per la postura e

la deambulazione, oltre che con tecniche correlate alla realtà virtuale; per la riabilitazione neuromotoria, come recuperare la perdita della deambulazione e come migliorarla associando metodiche comportamentistiche ABA; riflessioni sulla più efficace terapia della scoliosi; una parentesi sulla musicoterapia, anche effettuata con il sostegno della CAA.

Una riflessione finale ci sensibilizzerà alla "care" psicologica delle nostre ragazze, nell'ottica di promuovere una migliore qualità di vita per loro e i loro familiari.

PROVIDER

C.I.S.E.F. - Centro Internazionale di Studi e Formazione "Germana Gaslini"
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Certificazione ISO9001:2008



RESEARCH

Investigating MeCP2 and JNK3 reciprocal regulation in human cellular models for Rett syndromeAlari Valentina¹, Cogliati Francesca¹, Tiziana Borsello², Lidia Larizza¹, Silvia Russo¹¹Molecular Genetics Laborator- Istituto Auxologico Italiano I.R.C. C.S-Milano- Italy; ²Neuroscience Department IRCCS-Istituto Di Ricerche Farmacologiche Mario Negri - Milano -Italy.**Deficient purposeful use of forepaws in female mice modelling Rett syndrome**De Filippis, Bianca^a, Musto, Mattia^a, Altabella, Luisa^b, Romano, Emilia^a, Canese, Rossella^b, Laviola, Giovanni^a.^aBehavioural Neuroscience Section, Dept. Cell Biology & Neuroscience, Istituto Superiore di Sanità, Roma, Italy; ^bMolecular and Cellular Imaging Section, Dept. Cell Biology & Neuroscience, Istituto Superiore di Sanità, Roma, Italy.**GSK3-beta inhibitors: a promising strategy for the CDKL5 variant of Rett syndrome**

Fuchs Claudia, Rocchina Viggiano, Norma Fustini, Stefania Trazzi, Marianna De Franceschi, Renata Bartesaghi & Elisabetta Ciani

Department of Biomedical and Neuromotor Sciences, University of Bologna, Italy.

N-glycosylation pattern changes for brain NPP-5 in Mecp2-mutant murine models of Rett syndromeHayek Joussef¹, Alessio Cortelazzo^{1,2}, Roberto Guerranti², Cinzia Signorini³, Silvia Leoncini^{1,3}, Alessandra Pecorelli^{1,3}, Francesco Scalabrin^{4,5}, Michele Madonna⁴, Stefania Filosa^{4,6}, Cinzia Della Giovampaola⁷, Antonietta Capone⁷, Thierry Durand⁸, Cristiana Mirasole⁹, Lello Zolla⁹, Giuseppe Valacchi¹⁰, Lucia Ciccoli³, Jacky Guy¹¹, Maurizio D'Esposito^{4,12}, Claudio De Felice¹³¹ Child Neuropsychiatry Unit, University Hospital (AOUS), Policlinico "S. M. alle Scotte", Siena, Italy; ² Department of Medical Biotechnologies, University of Siena, Siena, Italy; ³ Department of Molecular and Developmental Medicine, University of Siena Via Aldo Moro, Siena, Italy; ⁴ IRCCS Neuromed, Pozzilli, Italy; ⁵ Department of Biochemistry, Biophysics, and General Pathology, 2nd University of Naples, Naples, Italy; ⁶ Institute of Biosciences and BioResources CNR, Naples, Italy; ⁷ Department of Life Science, University of Siena, Siena, Italy; ⁸ Institut des Biomolécules Max Mousseron (IBMM), UMR 5247, CNRS/UM1/UM2/ENSCM, BP 14491, 34093 Montpellier Cedex 5, France; ⁹ Department of Ecological and Biological Sciences, University of Tuscia, Viterbo, Italy; ¹⁰ Department of Life Sciences and Biotechnology, University of Ferrara, Ferrara, Italy; ¹¹ The Wellcome Trust Centre for Cell Biology, University of Edinburgh, Edinburgh, UK; ¹² Institute of Genetics and Biophysics "Adriano Buzzati Traverso" CNR, Naples, Italy; ¹³ Neonatal Intensive Care Unit, University Hospital, AOUS, Siena, Italy.**Improvement in Rett syndrome-like symptoms in Mecp2-deficient male and female mice after administration of a self-complementary AAV9 (scAAV9) construct expressing a codon-optimized Mecp2 transgene.**Matagne V^{1,2}, Villard L^{1,2}, Roux JC^{1,2}¹Inserm, UMR_S 910, Faculté de Médecine de La Timone, Marseille, France; ²Aix Marseille Université, GMGF, Marseille, France.**Mouse models of common Rett syndrome mutations recapitulate the severity seen in patients.**

Selfridge Jim, Kyla Brown, Sabine Lagger, John Connelly, Dina De Sousa, Alastair Kerr, Shaun Webb, Jacky Guy, Cara Merusi, Martha V. Koerner and Adrian Bird

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Construction of CNF1 autoassembling variants endowed with blood-brain barrier crossing ability: an innovative therapeutic approach for RTTTutino Maria Luisa¹, Gennaro Antonio Apuzzo¹, Andrea Colarusso¹, Alessia Fabbri², Ermenegilda Parrilli¹, Bianca De Filippis³, Laura Ricceri³, Neus Ferrer Miralles⁴, Giovanni Laviola³, and Carla Fiorentini²¹Dipartimento di Scienze Chimiche, Università di Napoli Federico II- tutino@unina.it; ²Dipartimento del Farmaco Istituto Superiore di Sanità, Viale Regina Elena, 00100 Roma; ³Dipartimento di Biologia Cellulare e Neuroscienze, Istituto Superiore di Sanità, Viale Regina Elena, 00100 Roma; ⁴Institut de Biociències i de Biomedicina, Departament de Genètica i de Microbiologia, and CIBER de Biotecnologia, Biomateriales y Nanomedicina (CIBER-BBN); Universitat Autònoma de Barcelona Bellaterra, 08193 Barcelona, Spain.

GENETICS

Investigation of mutation types in Rett syndrome in Denmark - Is long-time survival with MECP2 mutations connected to type of mutation?Bitten Schönewolf-Greulich & Anne-Marie Bisgaard
Centre for Rett syndrome, Department of Clinical Genetics - Kennedy Centre, Rigshospitalet, Copenhagen.**Genetic diagnosis of patients with overlapping clinic Rett-like by NGS**Brandi Tarrau NM¹, Gerotina Mora E², Vidal Falcó S², Ortez González CI³, García Cazorla A³, Gean-Molins E⁴, Pineda Marfa M², Armstrong Morón J¹.¹Department of Biochemistry Genetics&Rett, ²Neurology and Clinic Genetics, University Hospital Sant Joan de Déu, Barcelona, ³Sant Joan de Déu, Barcelona.**MECP2 Duplication: genetic and clinical study in Spanish patients**Falcó S Vidal¹, Blasco Pérez L¹, Gean Molins E², del Campo Casanelles M³, Català Cahis V⁴, Guillén E⁵, Lapunzina P⁶, Lloveras E⁷, Maortua H⁸, Martínez Castellano F⁹, Mesas Aróstegui A¹⁰, Nevado J⁶, Obón Ferrer M¹¹, Orellana Alonso C⁹, Plaja A¹², Roselló Piera M⁹, Tejada M I⁸, Tizzano E¹³, Armstrong Morón J¹⁴.¹Fundación San Juan de Dios, Barcelona. ²Unidad de consejo genético, Hospital Universitario San Juan de Dios, Barcelona. ³Pediatrics, Genetic Epidemiology, Hospital Valle Hebrón, Barcelona. ⁴Unidad de

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Our experience in genetic diagnosis of Rett syndrome by Next Generation Sequencing

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Familial Cases with MECP2 Mutations

Qingping Zhang, Ying Zhao, Xiaoying Zhang, Xiru Wu, Xinhua Bao

Department of Pediatrics, Peking University First Hospital, Beijing 100034, China.

Novel MECP2 mutations in Czech patients with Rett syndrome

Zahorakova Daniela¹, Alice Baxova², Vladimír Gregor³, Martina Langová³, Martin Magner¹, Zumrova Alena⁴, Pavel Martasek¹

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CLINICS

Nocturnal alterations in Rett syndrome: a polysomnographic study

Maria Esposito, Francesco Prencziano, Valentina Lanzara, Marco Carotenuto

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Recurrent fractures in a Rett patient treated with teriparatide

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The Influence of Mobility on Bone Status in Subjects with Rett Syndrome: a 10-Year Longitudinal Study

Caffarelli C., ¹M.D. Tomai Pitinca, ^{1V.} Francolini, ²J. Hayek, ^{1R.} Nuti, ^{1S.} Gonnelli

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Sleep disturbance in Rett Syndrome

Gagliardini G., M.Traverso, T. Giacomini, S. Uccella, M.Pintaudi, E.Veneselli

U.O. Neuropsichiatria Infantile, Istituto Giannina Gaslini.

Midazolam as treatment of recurrent seizures in a child with Rett Syndrome

Germano Michele

Neuropsichiatria Infantile, IRCCS "Casa Sollievo della Sofferenza", San Giovanni Rotondo (FG).

GH deficiency and growth retardation in Rett syndrome

Giacomini T¹, Pintaudi M¹, Mancardi MM¹, Traverso M¹, Gagliardini G¹, Uccella S¹, Napoli F², Di Iorgi N², Veneselli E¹

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Abnormal adipocyte-derived hormones in Rett syndrome mirroring chronic oxidative stress and inflammation: effects of ω-3 PUFAs

Hayek Joussef¹, Silvia Leoncini^{1,2}, Cinzia Signorini², Thierry Durand³, Jean-Marie Galano³, Camille Oger³, Jetty Chung-Yung Lee⁴, Gloria Zollo^{1,2}, Alessio Cortelazzo^{1,5}, Roberto Guerranti⁵, Lucia Ciccolì², Claudio De Felice⁶

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Abnormal visual evoked potentials, eye misalignment and visual acuity deficit in Rett syndrome: relationship with myelin oxidative damage

Hayek Joussef¹, Martina Zevolini², Bruno Natale², Cinzia Signorini³, Silvia Leoncini^{1,3}, Vincenzo Baranello⁴, Gloria Zollo^{1,3}, Lucia Ciccolì³, Jean-Marie Galano⁵, Camille Oger⁵, Thierry Durand⁵, Claudio De Felice⁶

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Abnormal erythrocyte morphology in CDKL5 Rett syndrome

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Biochemical characteristics of inflammation in CDKL5-Rett syndrome: plasma protein patterns and effects of ω -3 PUFAs

Hayek Joussef¹, Alessio Cortelazzo^{1,2}, Alessandro Armini³, Luca Bini³, Silvia Leoncini^{1,4}, Cinzia Signorini⁴, Roberto Guerranti², Lucia Ciccoli⁴, Claudio De Felice⁵

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Dietary habits in Rett syndrome

Hayek Joussef¹, Moraika Riggi², Francesca Cardinalli², Silvia Leoncini^{1,3}, Cinzia Signorini³, Alessio Cortelazzo^{1,4}, Gloria Zollo^{1,3}, Lucia Ciccoli³, Claudio De Felice⁵

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Oxidative stress in MECP2 duplication syndrome

Hayek Joussef¹, Cinzia Signorini², Silvia Leoncini^{1,2}, Rikke S. Møller^{3,4}, Gloria Zollo^{1,2}, Sabrina Buoni¹, Alessio Cortelazzo¹, Roberto Guerranti⁵, Thierry Durand⁶, Lucia Ciccoli², Maurizio D'Esposito^{7,8}, Kirstine Ravn⁹, Claudio De Felice¹⁰

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Unexplained sudden death in Rett syndrome: protective effect of ω -3 PUFAs

Hayek Joussef¹, Cinzia Signorini², Silvia Leoncini^{1,2}, Alessio Cortelazzo^{1,3}, Gloria Zollo^{1,2}, Thierry Durand⁴, Jean-Marie Galano⁴, Camille Oger⁴, Jetty Chung-Yung Lee⁵, Jean-Yves Le Guennec⁶, Silvia Maffei⁷, Alessandra Pecorelli^{1,2}, Giuseppe Valacchi⁸, Lucia Ciccoli², Claudio De Felice¹

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Clinical severity and multidimensional approach in Rett syndrome: two case reports

Traverso M¹, Gagliardini G.¹, Giacomini T¹, Uccella S.¹, Pintaudi M¹, Pedemonte M², Mancardi M.M.¹, Veneselli E.¹

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COGNITION

Water rehabilitation in Rett Syndrome.

Costagliola Romana

Neurophysiological and cognitive effects of Transcranial Direct Current Stimulation in Rett syndrome with chronic language impairments.

Fabio Rosa Angela^{*}, Antonio Gangemi^{*}, Tindara Capri^{*}, Alessandra Falzone^{*}

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Consistency of Choice Behavior in Rett syndrome

Fabio R.A.¹, Magaudda, C.¹, Falzone, A.M.¹, Capri, T.¹, Carrozza, C.¹, Towey, G.¹, Cedro, C.²

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Development of Clinical Guidelines for the Management of Communication in Individuals with Rett syndrome

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Comparing Social Reciprocity in Preserved Speech Variant and Typical Rett syndrome during the Early Years of Life

Townend Gillian S^a, Katrin D Bartl-Pokorny^b, Jeff Sigafoos^c, Leopold M G Curfs^a, Sven Bölte^d, Luise-Poustka^{e,f}, Christa Einspieler^b, Peter B Marschik^{b,d,g}

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