

# April 25-27, 2014 Bologna, Italy

**Department of Biomedical and Neuromotor Sciences  
University of Bologna, Physiology Building,  
Piazza di Porta San Donato 2, 40126 Bologna, Italy**

## *First day session*

**In the search of a therapy for the CDKL5 disease:  
joint efforts of the scientific community.**

## *Second day session*

**Characterization of the CDKL5 disease:  
international clinical network.**

## *Third day session*

**The family: how to help our children.  
Therapies for rehabilitation**

# Second International Meeting of the CDKL5 Associations April 25-27, 2014 - Bologna, Italy

**FRIDAY, APRIL 25, 2014**

Free medical examination by  
appointment  
Gobbi and Renieri

- 8:30-9:00 **REGISTRATION**
- 9:00-9:15 **Official opening of the meeting**  
Renata Bartesaghi, University of Bologna
- 9:15-9:30 **Emilia Romagna Area - Rare diseases**  
Elisa Rozzi, Matteo Volta - Italy
- 9:30-10:00 **Telethon and Patient - Organizations: synergistic actions for the progress of science**  
Francesca Sofia, Telethon Foundation

## **Session 1. Chairs: Renata Bartesaghi, Maurizio Giustetto**

- 10:00-10:30 **Modeling CDKL5 Disorder in Genetically Modified Mice**  
Judy Wang, University of Pennsylvania School of Medicine, Philadelphia - USA
- 10:30-11:00 **Rett syndrome-related genes share common pathogenic pathways**  
Maurizio Giustetto, Università di Torino, Torino - Italy
- 11:00-11:30 **Impaired cognitive function and hippocampal neurogenesis in a new model of CDKL5 Disorder**  
Elisabetta Ciani, Department of Biomedical and Neuromotor Sciences University of Bologna, Bologna - Italy

## **Session 2. Chair: Laura Calzà**

- 11:30-12:00 **Dendritic spine dynamics in cortical neurons of CDKL5 null mice**  
Tommaso Pizzorusso, Università di Firenze, Firenze - Italy
- 12:00-12:30 **Cerebellar function and networks in CDKL5 KO mouse**  
Laura Calzà, University of Bologna, Bologna - Italy
- 12:30-13:00 **Synaptic function of CDKL5**  
Zhi-Qi Xiong, Institute of Neuroscience and State Key Laboratory of Neuroscience, Shanghai Institutes for Biological Sciences, Chinese Academy of Sciences, Shanghai - China
- 13:00-14:30 **LUNCH TIME**

## **Session 3. Chairs: Giuseppe Gobbi, Pawel Stankiewicz**

- 14:30-15:00 **Recent gene therapy approaches in models of Rett syndrome**  
Stuart Cobb, University of Glasgow, Glasgow - UK
- 15:00-15:30 **GluD1 is a common altered player in neuronal differentiation from both MECP2-mutated and CDKL5-mutated iPS**  
Alessandra Renieri, Azienda Ospedaliera Universitaria Senese, Siena - Italy
- 15:30-16:00 **Synaptic synthesis, dephosphorylation and degradation: a novel paradigm for a developmentally regulated NMDA-dependent control of CDKL5**  
Nicoletta Landsberger, Department of Theoretical and Applied Sciences, University of Insubria Busto Arsizio (VA) - Italy
- 16:00-16:30 **A novel subcellular meeting point for MeCP2 and CDKL5: possible functional implications in normal and pathological conditions**  
Charlotte Kilstrup-Nielsen, Department of Theoretical and Applied Sciences, University of Insubria Busto Arsizio (VA) - Italy
- 16:30-17:00 **Mosaic exonic deletions and gene duplications of CDKL5 in males and females**  
Pawel Stankiewicz, Baylor College of Medicine, Houston TX- USA
- 17:00-17.30 **Science and pseudoscience today**  
Carlo Hanau, University of Modena and Reggio Emilia - Italy
- 17:30-18.00 **Nutraceutical: A new approach to drug discovery**  
Matteo Micucci, Roberta Budriesi, Department of Pharmacy and Biotechnology University of Bologna - Italy
- 18:00-18.30 **General discussion**
- 18:30-20:00 **WELCOME DRINK**

**Thanks to interpreters: Francesco Checchi, Michela Bertozzi, Elena Di Concilio**

## SATURDAY, APRIL 26, 2014

Free cranio sacral therapy  
by appointment  
Quirini

8:30-9:00 **REGISTRATION**

9:00-9:30 **Welcome**

### **Session 4. Chair: Silvia Russo**

- 9:30-10:00 **Molecular screening of CDKL5 in patients with early-onset seizures encephalopathies: the French experience**  
Christophe Philippe, Laboratoire de Génétique Médicale INSERM - France
- 10:00-10:30 **Behavioral phenotype in CDKL5 Syndrome: literature review and personal experience**  
Paola Visconti, Neuropsichiatria Infantile, Ospedale Maggiore, Bologna - Italy
- 10:30-11:00 **Diagnosing CDKL5: mutational screening and focus on large deletions involving CDKL5 and flanking regions**  
Silvia Russo, Istituto Auxologico Italiano, Cusano Milanino, Milano - Italy

### **Session 5. Chair: Renzo Guerrini**

- 11:00-11:30 **Analysis of phenotype in Rett Syndrome Networked Database: differences and similarities between MECP2 and CDKL5 patients.**  
Alessandra Renieri, Azienda Ospedaliera Universitaria Senese Siena - Italy
- 11:30-12:00 **Similarities and differences between Rett syndrome and CDKL5 disorder**  
Giorgio Pini, Tuscany Rett Center, Versilia Hospital, 55043 Lido di Camaiore - Italy
- 12:00-12:30 **CDKL5 gene related epileptic encephalopathy**  
Renzo Guerrini, Meyer Hospital, Firenze - Italy
- 13:00-14:00 **LUNCH TIME**

### **Session 6. Chair: Giuseppe Gobbi**

- 14:00- 14:30 **Molecular and Clinical Analysis of CDKL5 gene in patients with Rett-like features: The Greek experience**  
Stavroula Psoni, Choremio Research Laboratory, Agia Sophia Children's Hospital, Athens - Greece
- 14:30-15:00 **CDKL5 mutated patients in Spain**  
Judith Armstrong, Department of Biochemical Genetics&Rett, Hospital Sant Joan de Déu, Barcelona - Spain
- 15:00- 15:30 **Respiratory consequences of the CDKL5 gene mutation: A clinical-biochemical appraisal on patients with the early onset variant of Rett syndrome**  
Joussef Hayek, Neonatal Intensive Care Unit S. M. Le Scotte General Hospital, Siena - Italy
- 15:30- 16:00 **Respiratory and sleep disorders in patients with CDKL5 syndrome**  
Eveline Hagebeuk, Kinderneurologel Stichting Epilepsie Instellingen - Nederland
- 16:00- 16:30 **Oxidative post-transductional modifications are responsible for SR-BI loss in CDKL5 fibroblasts**  
Giuseppe Valacchi, Department of Life Science and Biotechnologies, University of Ferrara, Ferrara - Italy.

### **Session 7. Chair: Elisabetta Ciani**

- 16:30- 17:00 **Horizon 2020: European perspectives for research on Rare Diseases**  
Stefania Trazzi, ARIC, University of Bologna
- 17:00- 17:30 **The Italian Institute of Health: perspectives for research on Rare Diseases**  
Domenica Taruscio, Istituto superiore di sanità.
- 17:30- 18:00 **Rett syndrome and CDKL5 disorder: a common perspective**  
Lucia Dovigo, Rett Syndrome foundation Airett, Italy
- 18:00-18:30 **Panel discussion: Characterization of the CDKL5 disease**  
Panel group for discussion: Giuseppe Gobbi, Giorgio Pini, Christophe Philippe, Alessandra Renieri
- 18:30-19:00 **Panel discussion: In search of a therapy for the CDKL5 disease**  
Panel group for discussion: Elisabetta Ciani, Stuart Cobb, Maurizio Giustetto, Nicoletta Landsberger, Tommaso Pizzorusso
- 19:00-19:30 **CONCLUSION REMARKS**

**Thanks to interpreters: Sara Machiavelli, Donatella Baggio, Chiara Gandolfi**

**SUNDAY, APRIL 27, 2014**

**Free medical examination  
by appointment  
Guerrini and Russo**

**Free dental examination  
by appointment  
Piana**

8:30-9:00 **REGISTRATION**

**Session 8. Chair: Cosimo Ricciutello**

- 9:00-9:20 **Hydrotherapy, methods for a finalized intervention**  
Massimo Quaranta, Istruzione Superiore Don Lorenzo Milani Pertini, Grottaglie (TA) - Italy
- 9:20-9:40 **A parents experience of spider therapy in the UK**  
Carol-Anne Partridge, Chairperson of CDKL5 UK, London -UK
- 9:40-10:00 **A love that gives support: pranotherapy. The experience of Giorgia**  
Rosanna Balsamo, Centro olistico "Dal punto luce" Rho Milano - Italy. Laura Faedda, Milano - Italy
- 10:00-10:20 **Psychology and equestrian rehabilitation as help for rare genetic syndromes**  
Angela Zampa, Aiasport Bologna - Italy
- 10:20-10:40 **The Upledger cranio sacral technique for the CDKL5 disease**  
Patricia Quirini, Cranio sacral Academy of Trieste - Italy
- 10:40-11:00 **CDKL5 and speech therapy: a possible challenge**  
Atalia Tresoldi Ravaioli - Italy
- 11:00-11:20 **How to make more independent CDKL5 children**  
Maddalena Pentella - Italy
- 11:20-11:40 **Pet therapy: Communications and interactions between Children and Dogs**  
Angela Zampa, Aiasport Bologna - Italy
- 11:40-12:00 **Improving oral health in children with special needs**  
Gabriela Piana, Department of Biomedical and Neuromotor Sciences University of Bologna, Bologna - Italy
- 12:00-12:20 **Special education: Psychopedagogy**  
Carlo Hanau, President of FederAMrare - Italy
- 12:20-12:40 **Rehabilitation of children in Italy**  
Emanuela Defranceschi, and President of the Italian CDKL5 Association
- 12:40-13:00 **ABA method, dance therapy and others**  
Katheryn Elibri, President of the International Foundation for CDKL5
- 13:00-14:00 **LUNCH TIME**
- 14:00-14:20 **UICI Bologna, parents committee experience: together for our children**  
Daniele Epifani, UICI Regional Committee Coordinator - Italy
- 14:20-14:40 **The importance of early treatments and rehabilitative interventions: Lega del Filo d'Oro**  
Patrizia Ceccarani, Special Unit Director for deaf-blind sensorial multi-impaired - Lega del Filo d'Oro onlus - Italy

**Session 9. Chair: Giuseppe Gobbi**

- 14:40-15:00 **Italian CDKL5 Association, our commitment**  
Antonella Statello, Vice President of the Italian CDKL5 association, Castel S. Pietro T. (Bo) - Italy
- 15:00-15:20 **CDKL5 UK, our commitment**  
Adrian Partridge, CDKL5 UK, London - UK
- 15:20-15:40 **International Foundation for CDKL5, our commitment**  
Katheryn Elibri, President of the International Foundation for CDKL5 Research - USA
- 15:40-16:00 **L'albero di Greta, our commitment**  
Barbara Maggiorotto, President of the association l'albero di Greta
- 16:00-17:30 **MEETING OF THE CDKL5 ASSOCIATION EXECUTIVE, PERSPECTIVES FOR THE FUTURE**

*The entire conference will be simultaneously translated from English into Italian and from Italian into English thanks to qualified interpreters. During the congress there is the possibility to leave children in a play area equipped with a soft corner, appropriate equipment and professional graduated and experienced educators.*

*"Patch Adams" will be present for the enjoyment of children.*

**Thanks to interpreters: Marina Cantoni, Sara Machiavelli**

Booking for free medical, dental, cranio sacral examinations: email adress: [emydefranceschi@hotmail.com](mailto:emydefranceschi@hotmail.com)  
[www.cdl5.altervista.org](http://www.cdl5.altervista.org) - [www.cdkl5.org](http://www.cdkl5.org)