



# 4th International CDKL5 Congress 2-3 June 2017 - Rome

**A.Roma Lifestyle Hotel and Conferencing Center**

*Via Giorgio Zoega, 59 - 00164 Rome, Italy*

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**Patronage**



REGIONE  
LAZIO

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Assessorato alla Persona, Scuola e  
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## Partners



# Friday 2

**08.00/08.30** Welcome Coffee & Registration  
**08.30/09.30** Welcome address - *Antonino Caridi*  
CDKL5 in Italy - *Omar Fumagalli*  
Lou Lou Foundation - *Majid Jafar*  
Istituto Superiore Sanità - *Domenica Taruscio*  
Telethon Italia - *Anna Maria Cazzato*

**09.30/10.30** **BASIC DISCOVERY** - CHAIR - *Dr.T. Pizzorusso, PhD*  
John Rouse, PhD - *University of Dundee*  
*Recent advances in understanding the mode of action of the CDKL5 kinase*  
Maurizio Giustetto, PhD - *University of Turin*  
*Synaptic determinants of CDKL5 disorder: therapeutic effect of mGlu receptors modulation*  
Charlotte Kilstrup-Nielsen, PhD - *University of Insubria*  
*Pregnenolone - a novel drug-based therapies for CDKL5 disorder?*  
DISCUSSION

**10.30/11.30** **DISEASE MODELS** - CHAIR - *A. Muotri, PhD*  
David Wyllie, PhD - *University of Edinburgh*  
*Pathophysiology of the CDKL5 rat model*  
Douglas Coulter, PhD - *Children's Hospital of Philadelphia*  
*Hippocampal Circuit Phenotypes in an Experimental Model of CDKL5 Disorder*  
Alysson R. Muotri, PhD - *University of California San Diego*  
*Modeling CDKL5 syndrome with human organoids*  
DISCUSSION

**11.30/12.00** **Coffee Break**

**12.00/13.00** **NEW THERAPEUTIC DEVELOPMENTS** - CHAIR - *S. Cobb, PhD*  
Michael Green, PhD - *University of Massachusetts Medical School*  
*Pharmacological Reactivation of the Xi-Linked CDKL5 Gene as a Therapeutic Approach for CDKL5 Deficiency*  
Kyle Fink, PhD - *University of California, Davis Medical Center*  
*Development of TALE and CRISPR/Cas9 as a putative therapy for treatment or correction of genetically-linked CDKL5 Deficiency*  
Stefania Trazzi, PhD - *University of Bologna*  
*Pharmacological approaches to rescue brain development in CDKL5 disorder*  
DISCUSSION

**13.00/14.00** **Lunch & Poster Session**

**14.30/16.00**

**CDKL5 DISORDER** - CHAIR - *Tim Benke, MD, PhD*

*Maria Trivisano, MD, PhD - Bambin Gesù Hospital, Rome*

***Ganaxolone in children with CDKL5 gene-related epileptic encephalopathy: preliminary analysis from an open-label trial***

*Tim Benke, MD, PhD - University of Colorado, School of Medicine*

***Clinical phenotypes of CDKL5 disorder seen in the IFCR Centers of Excellence***

*Stefania Bigoni, MD. - University of Ferrara*

***CDKL5 disorder and Hanefeld RTT variant: clinical, genetic and autonomic picture of 12 cases belonging to different European countries***

*Domenica Battaglia, MD, PhD - Policlinico Gemelli Hospital, Rome*

***Electroclinical phenotype in children with CDKL5-gene related disorders***

*DISCUSSION*

**16.00/16.30**

**Coffee Break**

**16.30/18.00**

**PANEL ON INDUSTRY PROSPECTIVE** - CHAIR - *AMICUS Therapeutics*

*AMICUS Therapeutics*

*PTC Bio*

*Marinus Pharmaceuticals*

*Ultragenyx*

*TAKEDA*

*ROCHE*

**20.00**

**SOCIAL DINNER**

# Saturday 3

**08.30/09.00** Coffee & registration

**09.00/09.30** Summary of 2 June for Families - *Elisabetta Ciani PhD*

**09.30/11.00 CARE GIVERS**

*Dr. Giorgia Gugelmo - Dietician*

***Ketogenic Diet. Family Routes: looking for new equilibriums***

*Dr. Tiziana Gobetti - Psychologist - Fondazione Santa Lucia IRCCS*

***Family roads: finding new balances***

*Maria Mangeruga - Fondazione Santa Lucia IRCCS - Alternative Communication*

***How to manage communication, interrelation and behavioural aspects: augmentative alternative communication.***

*DISCUSSION*

**11.00/11.30 Coffee Break**

**11.30/13.00** *Dr. Lorenza Citeroni - Physiatrist*

***Functional bandages***

*De Souza Oliveira Francielly*

***Therapies with dynamic proprioceptive orthoses***

*Belenguer Benitez Charo and Escobar Torres Maria Lourdes*

***MITAF (Integral Method of Functional AQUATIC Work)***

***Aquatic programme adapted to people with a disability***

*DISCUSSION*

**13.00/14.00 Lunch**

**14.30/15.30** **CDKL5 Register** – *Umberto Pasqualetto* - Association CDKL5 Insieme verso la cura  
*Michela Quintiliani Phd* - Policinico Gemelli Hospital Rome  
*Maria Luigia Gambardella Phd* - Policinico Gemelli Hospital Rome  
**International CDKL5 Registry**  
*Ashley R. Winslow, Phd* - Orphan Disease Center & Lou Lou Foundation, and Pulse Infoframe  
*Tamara Howard, Phd* - Pulse Infoframe  
*DISCUSSION*

**15.30/ 16.00** **Coffe Break**

**16.00/17.30** **Family Associations**  
**European Families Associarions**  
*Carolanne Partridge (CDKL5 UK) – A road to diagnosis and beyond*  
*Sandra Thorpe Bione - Doctor Axel Panzer (CDKL5 Deutschland)*  
**Alliance against CDKL5**  
*Pam Baauw (CDKL5 Netherland ) - Chair of the CDKL5 Netherlands Community*  
*Alexandra Mikulec (CDKL5 Slovakia)*  
*Katheryn Elibri (International Foundation for CDKL5 Reserch - USA)*  
  
Other associations: Spain, Russia, Canada, France, Ireland, Belgium.

**17.30/18.00** **Conclusion**

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## Scientific Committee

*Elisabetta Ciani, PhD*  
Associate Professor of Physiology  
Dept. Biomedical and Neuromotor Sciences  
University of Bologna

*Ashley R. Winslow, PhD*  
CSO, Loulou Foundation  
Sr. Director, CDKL5 Program of Excellence &  
Neurogenetics  
Orphan Disease Center (Univ. Pennsylvania)

## Friends



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