

# 7<sup>TH</sup> WORLD CONGRESS ON RETT SYNDROME

## TRANSLATIONAL & PRE-CLINICAL RESEARCH SYMPOSIUM

### JUNE 24, 2012

SUNDAY, JUNE 24, 2012	
7:30 – 8:45am	<b>BREAKFAST – Pontalba Reception Area</b>
8:45-9:00am	<b>Opening Remarks / Meeting &amp; Session Overview – Jeffrey Neul, MD, PhD Baylor College of Medicine</b>
<b>I. PRE-CLINICAL TRIALS IN RETT SYNDROME</b> <b>Chair: Laurent Villard, PhD, Inserm UMR</b>  <i>Pontalba</i> <b>9:00-11:00 am</b>	
9:00 – 9:20am	<i>Deep brain stimulation in the striatum improves motor function, enhances weight gain and extends longevity in the MeCP2null/y mouse model of Rett Syndrome</i> James Leiter, MD, Dartmouth College
9:20 – 9:40am	<i>Modification of Mecp2 dosage alters the axonal transport: from molecular mechanisms to pharmacological treatment</i> Jean-Christophe Roux, PhD Inserm UMR
9:40 – 10:00am	<i>Chronic treatment with high dose PEGylated Insulin like growth factor I worsens a metabolic syndrome and shortens lifespan in Mecp2 null mice</i> Jeffrey Neul, MD, PhD, Baylor College of Medicine, Jan and Dan Duncan's Neurological Research Institute at Texas Children's Hospital
10:00 – 10:20am	<i>The sphingosine-1 phosphate receptor modulator fingolimod increases BDNF levels and improves symptoms of mice lacking Mecp2</i> Rubén Deogracias, PhD, Biozentrum, University of Basel
10:20 – 10:40am	<i>MeCP2 Gene Therapy for Rett Syndrome: Proof of Concept and Translational Obstacles</i> Steven Gray, PhD, University of North Carolina at Chapel Hill
10:40 – 11:00am	<i>Sarizotan, a full serotonin 1a receptor agonist, dopamine D2-like partial agonist effectively treats respiratory disorders in mouse models of Rett syndrome</i> John Bissonnette, MD, Oregon Health and Science University
11:00 – 11:15am	<b>MORNING BREAK – Pontalba Reception Area</b>
<b>II. OUTCOME MEASURES IN RETT SYNDROME</b> <b>Chair: Jeffrey Neul, MD, PhD, Baylor College of Medicine, Jan and Dan Duncan's Neurological Research Institute at Texas Children's Hospital</b> <i>Pontalba</i> <b>11:15-1:15pm</b>	
11:15 – 11:35am	<i>Pupillometry in Pediatric Respiratory and Autonomic Disorders: Quantitative Evidence of Autonomic Nervous System Dysregulation</i> Pallavi Patwari, MD, Northwestern University's Feinberg School of Medicine, Ann & Robert H. Lurie Children's Hospital of Chicago
11:35 – 11:55am	<i>Infrared thermal analysis and autonomic function in Rett syndrome</i> Frank Symons, PhD, University of Minnesota
11:55 – 12:15pm	<i>Physical activity in Rett syndrome</i> Jenny Downs, PhD, Telethon Institute for Child Health Research, University of Western Australia and School of Physiotherapy, Curtin University
12:15 – 12:35pm	<i>Biomarkers of cortical function in Rett Syndrome: characterization of anomalous brain activity and testing efficacy of pharmacological treatment with IGF1</i> Geneva DeGregorio, Children's Hospital Boston

12:35 – 12:55pm	<i>Assessment of cognitive functions in Rett syndrome: secrets of their eyes</i>
	Aleksandra Djukic, MD, PhD, Montefiore Medical Center, Albert Einstein College of Medicine
12:55 – 1:15pm	<i>Behavioral abnormalities in Rett syndrome: Overview and development of a rating scale</i>
	Walter Kaufmann, MD, Children's Hospital Boston, Harvard Medical School
1:30 – 2:30pm	<b>LUNCH &amp; POSTER SESSION – <i>Vieux Carre</i></b>
<b>III. CLINICAL TRIALS IN RETT SYNDROME</b> Chair: Peter Huppke, MD, University of Göttingen <i>Pontalba</i> 2:30 – 4:30pm	
2:30 – 2:50pm	<i>Initial study of rh-IGF1 (mecasermin [DNA] injection) for treatment of Rett syndrome and development of Rett-specific novel biomarkers of cortical and autonomic function</i>
	Eugenia Ho, MD, Children's Hospital Boston
2:50 – 3:10pm	<i>An update on the clinical trial with desipramin</i>
	Josette Mancini, MD, Children's Hospital Timone, Université de la Méditerranée
3:10 – 3:30pm	<i>TBD</i>
	Nadia Bahi-Buisson, MD, PhD, Necker Enfants Malades University Hospital, University Paris V Descartes
3:30 – 3:50am	<i>Rationale for the use of dextromethorphan and results of an open label dose effect study in RTT</i>
	Sakkubai Naidu, MD, Johns Hopkins University, Hugo Moser Research Institute at Kennedy Krieger, Inc
3:50– 4:30pm	<b>Discussion &amp; Wrap-Up – Round table discussion on challenges in clinical trials in Rett syndrome</b>
4:30pm	<b>Close Program</b>