

International Tuberous Sclerosis Complex (TSC) Conference: From DNA to Therapeutics

Wednesday, September 23, 2009

12:00 pm – 9:00 pm Registration Open

Dinner on your own

Opening Session I: An Overview of TSC and the mTOR Pathway Chair: Vicky Whittmore, PhD

6:30 pm – 7:30 pm Opening Reception

7:30 pm – 9:00 pm Welcome and Introductions – Kari Luther Carlson, President & CEO, and Celia Mastbaum, Board Chair, Tuberous Sclerosis Alliance

Pathological and Therapeutic Implications of mTOR Misregulation in TSC
Brendan Manning, PhD
Harvard School of Public Health, Boston, MA

Thursday, September 24, 2009

7:00 am – 10:00 pm Registration Open

7:30 am – 8:30 am Breakfast

Session I: Models and Mechanisms of TSC Pathology: Non-CNS Session Chair: David Kwiatkowski, MD, PhD

8:30 am – 8:40 am Parent or Individual with TSC

8:40 am – 9:40 am mTOR Signaling and Cancer
John Blenis, PhD
Harvard Medical School, Boston, MA

9:40 am – 10:05 am Function of the TSC-mTOR Pathway in Energy Balance
Kun-Liang Guan, PhD
University of California, San Diego

10:05 am – 10:30 am Mechanisms and Therapeutic Implications of AMPK Regulation of mTORC1 Signaling
Reuben Shaw, PhD
Salk Institute, La Jolla, CA

10:30 am – 10:45 am Break

10:45 am – 11:05 am Characterization of mTORC2 Regulation Through Rictor Phosphorylation: Identification of an IRS1-independent Feedback Mechanism
Christian Dibble
Harvard School of Public Health, Boston, MA

11:05 am – 11:25 am Defects in Cell Polarity Underlie TSC-and ADPKD-associated Cystogenesis
Cleo Bonnet, PhD
Cardiff University, Cardiff, UK

11:25 am – 11:45 am *TSC2*^{+/-} Mice Develop Tumors in Multiple Sites Due to the Activation of the Mesenchymal-specific Gene *Hmga2*
Patrick Geraghty, PhD
Columbia University, New York, NY

- 11:45 am – 12:05 pm An Epigenetic Event May Underlie the TSC Features in TSC2 ASM Cells
Elena Lesma, PhD
Universita degli Studi di Milano, Milano, Italy
- 12:05 pm – 12:25 pm TSC Missense Variants and Their Role in Disease
Rosemary Ekong, PhD
University College London
- 12:25 pm – 1:30 pm Lunch

Session II: Models and Mechanisms of TSC Pathology: CNS
Session Chair: Brenda Porter, MD, PhD

- 1:30 pm – 1:40 pm Parent or individual with TSC
- 1:40 pm – 2:40 pm Mouse Models as Translational Tools to Discover Treatments for Autism Spectrum Disorders: Focus on Rapamycin
Luis Parada, PhD
University of Texas Southwestern Medical School, Dallas, TX
- 2:40 pm – 3:05 pm Axonal Connectivity in Tuberous Sclerosis Complex
Mustafa Sahin, MD, PhD
Boston Children's Hospital, Boston, MA
- 3:05 pm – 3:30 pm Context-dependent Functions of PI3K Signaling in the Nervous System
Suzanne Baker, PhD
St. Jude's Children's Hospital, Memphis, TN
- 3:30 pm – 3:45 pm Break
- 3:45 pm – 4:05 pm Defects of Neural Development and Cell Size in a Zebrafish Model of Tuberous Sclerosis Complex
Kevin Ess, MD, PhD
Vanderbilt University, Nashville, TN
- 4:05 pm – 4:30 pm Glutamate transport and potassium uptake are impaired in astrocytes in human epileptic tuberous sclerosis tissue
Guy McKhann, MD
Columbia University, New York, NY
- 4:30 pm – 4:50 pm Regulable Loss of TSC1 in Neural Progenitor Cells Yields a Model of Tuberous Sclerosis Complex (TSC) with Giant Cells and Other Features Replicating Cortical Tubers
June Goto, PhD
Brigham and Women's Hospital, Boston, MA
- 4:50 pm – 5:10 pm The Generation and Characterization of a Rheb Knock Out Mouse
Susan Goorden, MSc
Erasmus Medical Center, Rotterdam, the Netherlands

Poster Session

- 5:30 pm – 7:30 pm Wine & Cheese Reception and Poster Session
- 7:30 pm – 9:30 pm Dinner

Friday, September 25, 2009

7:00 am – 7:00 pm Registration Open

7:30 am – 8:30 am Breakfast

Session III: Brain Development, Epileptogenesis and Neurocognition Session Chair: Joyce Wu, MD

8:30 am – 8:40 am Parent or individual with TSC

8:40 am – 9:40 am TBD
Elizabeth Thiele, MD, PhD
Massachusetts General Hospital, Boston, MA

9:40 am – 10:05 am The Neurodevelopmental Pathology of a Mouse Model in TSC
Michael Gambello, MD, PhD
University of Texas Health Science Center, Houston, TX

10:05 am – 10:30 am Designing Rational Therapies for Epilepsy in TSC
Michael Wong, MD, PhD
Washington University, St. Louis, MO

10:30 am – 10:45 am Break

10:45 am – 11:10 am Molecular, Synaptic, and Behavioral Abnormalities in Dominant/Negative TSC2 Mice
Eric Klann, PhD
New York University, New York, NY

11:10 am – 11:30 am Effects of Conditionally Inactivating *Tsc1* in the Thalamus During Development in a Mouse Model of Tuberous Sclerosis
Mark Zervas, PhD
Brown University, Providence, RI

11:30 am – 11:55 am TBD
Howard Weiner, MD
New York University, New York, NY

11:55 am – 12:20 pm Translational Neuroscience in TSC- Are We at Risk of Getting Lost Between the Bench and the Bedside?
Petrus de Vries, MD, PhD
University of Cambridge, Cambridge, UK

12:20 pm – 1:30 pm Lunch

Session IV: Current and Future Therapeutic Options for TSC and Related Disorders Session Chair: Aris Astrinidis, PhD

1:30 pm – 1:40 pm Parent or individual with TSC

1:40 pm – 2:40 pm Treating Tuberous Sclerosis: Understanding Its Biology Strengthens Our Hand
Julian Sampson, MB
Cardiff University, Cardiff, UK

2:40 pm – 3:05 pm Therapeutic Options and Biomarkers for TSC Related Tumors: Rapamycin, VEGF-D, and Angiogenesis Inhibitors
Sandra Dabora, MD, PhD
Brigham & Women's Hospital, Boston, MA

- 3:05 pm – 3:30 pm TSC Renal Disease: Novel Features and Implications
John Bissler, MD
Cincinnati Children's Hospital Medical Center, Cincinnati, OH
- 3:30 pm – 3:45 pm Break
- 3:45 pm – 4:05 pm TBD
David Franz, MD
Cincinnati Children's Hospital Medical Center, Cincinnati, OH
- 4:05 pm – 4:30 pm Epilepsy in TSC: How Early Can We Treat It?
Sergiusz Jozwiak, MD, PhD
Children's Hospital, Warsaw, Poland
- 4:30 pm – 4:50 pm Optical Coherence Tomography in Patients Taking Vigabatrin: Exploring New Methods to Assess Visual Field Loss
Lisa Clayton, BSc
University College London, London, UK
- 4:50 pm – 5:10 pm Cellular Targets of Rapamycin in a Xenograft Model of TSC Skin Tumors
Rajesh Thangapazham, PhD
Uniformed Services University of the Health Sciences, Bethesda, MD
- 7:00 pm – 9:00 pm Conference Dinner

Saturday, September 26, 2009

- 7:00 am – 12:00 pm Registration Open
- 7:30 am – 8:30 am Breakfast

Session V: Does Aberrant mTORC1 Signaling Explain Everything? Session Chair: Cheryl Walker, PhD

- 8:30 am – 8:40 am Parent or individual with TSC
- 8:40 am – 9:40 am Tuberosclerosis and LAM: Pathogenic Mechanisms
Elizabeth Petri Henske, MD
Brigham & Women's Hospital, Boston, MA
- 9:40 am – 10:05 am Pathogenesis of TSC through Epithelial Mesenchymal Transition (EMT)
Raymond Yeung, MD
University of Washington, Seattle, WA
- 10:05 am – 10:30 am Regulation of Signal Transducer and Activator of Transcription-1 (STAT1) by Mammalian Target of Rapamycin
Arnold Kristof, MD
McGill University, Montreal, Quebec, Canada
- 10:30 am – 10:45 am Break
- 10:45 am – 11:05 am 4-Phenylbutyric Acid: An AMPK Agonist and mTORC1 Inhibitor
Lawrence Quilliam, PhD
Indiana University School of Medicine, Indianapolis, IN
- 11:05 am – 11:25 am Pharmacologic Inhibition of Polo-like Kinase 1 (PLK1) Decreases the Survival of Hamartin and Tuberin Deficient Cells
Aristotelis Astrinidis, PhD
Drexel University College of Medicine, Philadelphia, PA
- 11:25 am – 11:55 am TACC3-TSC2 Maintain Nuclear Envelope Structure and Control Cell Division
Laia Gomez
Catalan Institute of Oncology, Barcelona, Spain

12:00 pm

Conference Adjourns