2017 International Research Conference on TSC and LAM: Innovating Through Partnerships

June 22  Day One, Thursday

2:00-6:30 p.m.  Welcome and Plenary session 1
Moderator: Steven L. Roberds, PhD, Tuberous Sclerosis Alliance

- 2:00-2:15 TSC/LAM patient or family member
- 2:15-2:45 Brendan Manning, PhD, Harvard T.H. Chan School of Public Health – Targeting anabolic balance downstream of mTORC1 to treat neoplastic lesions in TSC and LAM
- 2:45-3:15 Maikel Peppelenbosh, PhD, Erasmus Medical Center – Kinome profiling identifies PAK2 as a novel target in tuberous sclerosis complex treatment
- 3:15-3:30 Rebecca Ihrie, PhD, Vanderbilt University School of Medicine – Differential mTORc1 activity in neural stem cell subdomains leads to location specific tumor development in models of TSC
- 3:30-4:00 Kimberly Raab-Graham, PhD, Wake Forest School of Medicine – Parkinson protein 7 links aberrant calcium dynamics and glutamate receptor trafficking to a model of TSC

4:00-4:30 BREAK (30 min)

Moderator: Francis X. McCormack, MD, University of Cincinnati

- 4:30-5:00 Emily Osterweil, PhD, University of Edinburgh – mRNA and translation at synapses regulated by mTOR
- 5:00-5:15 Sungho Won, PhD, Seoul National University – A LAM Genome Wide Association Study (GWAS) implicates NR2F2 in LAM pathogenesis
- 5:15-5:30 Emmanuelle Logette, PhD, Association sclérose tubéreuse de Bourneville – The TSC burdens in France for patients and caregivers
- 5:30-5:45 Yun-Jung Chiang, PhD, Academia Sinica – CBAP critically regulates Akt-dependent inhibition of Rheb-GAP activity of TSC2 to promote Rheb-mTORC1 signaling axis and tumorigenesis
- 5:45-6:00 Jane Yu, PhD, University of Cincinnati College of Medicine – Tuberin negatively regulates the expression and function of prostaglandin receptors via Rheb
- 6:00-6:15 Helen Bateup, PhD, University of California, Berkeley – Second hit mutations generate cortical tuber-like cells in human neuronal models of tuberous sclerosis
- 6:15-6:30 Chris Ankney, Loyola University Chicago – TCR transgenic, gp100 reactive T cells to target lymphangioleiomyomatosis

7:00-9:00 p.m.  Dinner and Keynote Speaker:
7:00-7:10 Opening Remarks by Kari Luther Rosbeck and Sue Sherman
7:10-7:25 Dr. Walter Koroshetz, Director, National Institute of Neurological Disorders and Stroke
7:25-7:40 Dr. James Kiley, Director, Division of Lung Diseases, National Heart, Lung, and Blood Institute

8:00-9:00 Keynote Address by David Sabatini, MD PhD, Massachusetts Institute of Technology – Impact of basic research on advances in TSC and LAM clinical care
June 23

Day Two, Friday

8:30 a.m. to Noon

Plenary session 2
Moderator: Raymond S. Yeung, MD, University of Washington

- 8:30-8:45 TSC/LAM patient or family member
- 8:45-9:15 Elizabeth Thiele, MD PhD, Harvard Medical School – CBD effects on epilepsy in TSC and Lennox-Gastaut Syndrome
- 9:15-9:45 Vera Krymskaya, PhD MBA, University of Pennsylvania – Pulmonary lymphangioleiomyomatosis (LAM): from hallmarks of monogenic malignancy towards mechanism-driven hallmark-targeting therapies
- 9:45-10:00 Andrew Tee, PhD, Cardiff University – Identifying novel mechanisms and drug targets that induce a selective cytotoxic effect in model systems of tuberous sclerosis
- 10:00-10:15 Hilaire Lam, PhD, Brigham and Women’s Hospital – miR-21 inhibits the tumor suppressor argininosuccinate synthase 1 (ASS1) to ensure aspartate availability for pyrimidine biosynthesis in mTORC1 hyperactive cells

10:15-10:45 BREAK (30 min)

- 10:45-11:15 Jeanine D’Armiento, MD PhD, Columbia University – HMGA2 an absolute requirement for TSC tumorigenesis: an alternative therapy for tuberous sclerosis through an mTOR independent pathway
- 11:15-11:45 Darcy Krueger, MD PhD, Cincinnati Children’s Hospital Medical Center – Targeting mTOR to improve neurocognition in patients with tuberous sclerosis
- 11:45-12:00 Loren Leclezio, University of Cape Town – Multivariate data analysis identifies natural clusters of tuberous sclerosis complex-associated neuropsychiatric disorders (TAND)

Noon-1:00 p.m.
Lunch provided (1 hr)

1:00-3:00 p.m.
1:00-2:30 Breakout working groups (90 min)

- Clinical
- Genetics
- Signaling and drug targets

2:30-3:00 BREAK (30 min)

3:00-5:00 p.m.
Plenary session 3
Moderator: Ajay Gupta, MD, Cleveland Clinic

- 3:00-3:30 Mustafa Sahin, MD PhD, Harvard Medical School – TACERN biomarker studies
- 3:30-4:00 Shafali Jeste, UCLA David Geffen School of Medicine – Timing and mechanisms of atypical development in tuberous sclerosis complex
- 4:00-4:30 Elizabeth Henske, MD, Harvard Medical School – Achilles’ heel: therapeutic targeting of metabolic vulnerabilities in lymphangioleiomyomatosis
- 4:30-5:00 Yonghao Yu, PhD, University of Texas Southwestern Medical Center – Navigating downstream of mTORC1: a quantitative proteomic approach

5:00-7:00 p.m.
Poster session and reception (2 hours)
June 24  

**Day Three, Saturday**

8:30 a.m.-Noon  

**Plenary session 4**  
*Moderator: Lisa R. Young, MD PhD, Vanderbilt University*

- 8:30-8:45 TSC/LAM patient or family member
- 8:45-9:15 Ravi Jagasia, PhD, F. Hoffmann-La Roche – mTORC1 controls neurodevelopment and synaptic plasticity in a time-dependent manner in cell models of tuberous sclerosis
- 9:15-9:45 Martina Bebin, MD MPA, University of Alabama Birmingham – The role of EEG as a biomarker to predict impending epilepsy in seizure-naive tuberous sclerosis complex infants
- 9:45-10:00 Michael Gambello, MD PhD, Emory University School of Medicine – Metabolomic studies of TSC mouse model brains uncover major changes in the methylation pathway – implications for neuropathology and treatment
- 10:00-10:15 You Feng, PhD, Brigham and Women’s Hospital – Targeting secreted lysophospholipase D/autotaxin in tuberous sclerosis complex (TSC) and lymphangioleiomyomatosis (LAM)

10:15-10:45 BREAK (30 min)

- 10:45-11:15 David Kwiatkowski, MD PhD, Harvard Medical School – Molecular studies in EPISTOP
- 11:15-11:45 Sergiusz Jóźwiak, MD PhD, Children’s Memorial Health Institute – EPISTOP project – data from current status
- 11:45-12:00 Caroline LePoole, PhD, Loyola University Chicago – Opportunities to treat TSC-deficient tumors by GD3 CAR-T cells

Noon-1:00 p.m.  

Lunch provided (1 hr)

1:00 – 3:00 p.m.  

**Plenary session 5**  
*Moderator: Hope Northrup, MD, The University of Texas Health Science Center at Houston*

- 1:00-1:30 Cheryl Walker, PhD, Baylor College of Medicine – Regulation of peroxisome homeostasis by AMPK, ATM and mTOR kinases
- 1:30-2:00 John Blenis, PhD, Weill Cornell Medical College – mTORC1/S6K1: regulation of cell metabolism
- 2:00-2:15 Charilaos Filippakis, PhD, Brigham and Women’s Hospital – Lysosomal regulation of cholesterol homeostasis in tuberous sclerosis complex is mediated via NPC1 and LDL-R
- 2:15-2:30 David Neal Franz, MD, Cincinnati Children’s Hospital – Sustained seizure reduction with adjunctive everolimus for treatment-refractory seizures associated with tuberous sclerosis complex (TSC): Long-term results from the phase 3 EXIST-3 study

2:30-3:00 BREAK (30 min)

3:00-5:00 p.m.  

**Joint session**  
*Moderator: Elizabeth Henske, MD, Harvard Medical School*

- 3:00-4:30 Joint session with individuals and families affected by TSC and/or LAM
- 4:30-5:00 Conference close
### 2017 International Research Conference on TSC and LAM Steering Committee

Co-Chairs: Francis X. McCormack, MD, Univ. of Cincinnati and Steven L. Roberds, PhD, TS Alliance

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