

# INTERNATIONAL TSC RESEARCH CONFERENCE 3-5 NOVEMBER'16



Navigating TSC
Science to Therapies

LISBON- PORTUGAL CHAMPALIMAUD FOUNDATION

## PROGRAM

#### Thursday, Nov 3

10:00	11:00	Transfer to Champalimaud Foundation
11:00	13:00	Registration and lunch
13:00	13:10	Opening
SESSION 1		Navigating from Genes to Cells Chair: Julian Sampson, MD, Prof.
13:15	13:35	Differential mTOR Pathway Signaling in TSC Peter B. Crino, MD, Ph.D
13:40	14:00	Characterization of a tsc2 zebrafish model of Tuberous Sclerosis Complex Chloë Scheldeman, Ph.D Scholar
14:05	14:25	Signaling through TSC1 and TSC2: more than mTOR Kathrin Thedieck, MD, Prof.
14:25	14:45	Cell biology of TSC-deficiency in neurons Alessia Di Nardo, MD, Ph.D
14:45	15:05	Shaping different cilia for different cell types  Mónica Bettencourt Dias, Ph.D
15:10	15:30	Coffee break
15:35	15:55	Epilepsy in TSC: from mice to trials  Ype Elgersma, MD, Prof.
16:00	16:20	Autism and TSC: mechanisms and insights into therapeutic time windows Peter Tsai, MD, Ph.D
16:25	16:45	Understanding the mechanisms of cortical tuber-induced seizures  Angélique Bordey, Ph.D
16:50	17:10	EsTUpt: What is the TSC reality in Portugal?  Ana Escoval, Prof and Carla Nunes, Prof.
17:15	17:30	Whole exome sequencing identifies TSC1/TSC2 biallelic loss as the primary and sufficient driver event for renal angiomyolipoma development Krinio Giannikou, Ph.D
17:30	17:45	mTORC1 inhibition promotes epithelial differentiation and inhibits cyst/spheroid growth of TSC2-deficient renal cystic epithelial cells in 3-dimensional culture Brian J. Siroky, Ph.D
17:45	18:00	A novel role of tuberin in controlling ROS generation in Patients with TSC Samy Lewiz-Habib, Ph.D
18:00	18:30	Transfer to the Marriott
19:30	21:30	Dinner at the Marriott
21:00	24:00	Munich Room - space for networking Floor -1













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### Friday, Nov 4

6:30	7:30	Meet the Experts at Breakfast
7:45		Transfer to Champalimaud Foundation
SESS	ION 2	Navigating from Cells to Disease Chair: Petrus J de Vries, Prof.
8:30	9:00	Genetics of TSC: Update and Applications in Clinical Practice Julian Sampson, MD, Prof.
9:05	9:25	mTOR Inhibitors for the treatment of epilepsy David N. Franz, MD
9:30	9:50	Towards differentiating TSC children with and without autism using regional cortical connectivity  Andrew C. Papanicolau, Ph.D
9:55	10:15	Can we find natural clusters of Tuberous Sclerosis Complex Associated Neuropsychiatric Disorders? A Pilot feasibility study Loren Leclezio, MSc, Med (neurosci), Ph.D Scholar
10:15	10:30	Resting-state neurophysiological activity in young people with TSC vs. young people with idiopathic ASD, ADHD and ASD+ADHD Elizabeth Shephard, MD, Ph.D
10:30	10:45	Tuberous Sclerosis Complex neuropathology requires glutamate-cysteine ligase Jacek Jaworski, Ph.D
10:45	11:15	Coffee break
11:10	11:30	Microstructures at ultrahigh MRI fields for early detection of neuroplasticity Noam Shemesh, Ph.D
11:35	11:55	Imaging evaluation of Tuberous Sclerosis Complex: Recent advances and future directions Asim F. Choudhri, MD
12:00	12:20	Beyond Rapalogs: Achieving a Cytocidal Response in LAM and TSC Associated Tumors Elizabeth P. Henske, MD
12:25	12:45	TSC renal cystic disease & Final analysis of Exist 2  John J. Bissler, MD
12:45	13:45	Lunch
SESS	ION 3	Navigating Disease to Optimum Health Chair: Paolo Curatolo, MD
13:45	14:05	Neuroimaging of Tuberous Sclerosis Complex Simon K. Warfield, Ph.D
14:10	14:30	Long-term follow-up of TSC patients with epilepsy treated preventatively Sergiusz Józwiak, MD, Ph.D
14:35	14:55	Tuberous Sclerosis Complex: Cannabanoids in the treatment of epilepsy Elizabeth A. Thiele, MD, Ph.D













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15:00	15:20	Chloroquine-dependent regulation of cholesterol homeostasis in tuberous sclerosis complex-deficient cells Harilaos Filippakis, Ph.D
15:20	15:40	Topical Rapamycin to Erase Facial Angiofibromas in TSC – Multicenter Evaluation of Novel Therapy (TREATMENT Trial)  Mary Kay Koenig, MD
15:40	16:00	Molecular analysis of TSC1 and TSC2 genes in 174 foetuses following the prenatal detection of cardiac tumours at ultrasound examination  Marie Claire Malinge, MD
16:00	17:30	Poster session at the Foundation
17:30		Transfer to Marriot
19:00		Transfer to Gala Dinner
19:30	22:00	Gala Dinner
22:00	24:00	Munich Room - space for networking Floor -1

### Saturday, Nov 5

6:45 7:30 Meet the Experts at breakfast

7:45		Transfer to Champalimaud Foundation
SESSION 4		Navigating Implementation of Care Chair: Darcy A. Krueger, MD, Ph.D
8:30	8:50	Visual Health in Tuberous Sclerosis Complex Lauren Ditta, MD
8:55	9:15	Cutaneous Lesions of Tuberous Sclerosis: Advances in Pathogenesis and Management Teresa S. Wright, MD, FAAD, FAAP
9:20	9:40	The Heart in Tuberous Sclerosis Thomas M. Yohannan MD, FACC
9:45	10:05	Hypertension in TSC: should we screen?  Djalila Mekahli, MD, Ph.D
10:10	10:30	End stage renal disease in TSC: can it be avoided? Peter Janssens, MD
10:30	10:50	Coffee break
10:50	11:20	Navigating Behavioral Challenges in Tuberous Sclerosis Complex  Tanjala T. Gipson, MD
11:30	11:40	Burden of Disease and Challenges to Implementation of Care Anna C. Jansen, MD, Ph.D
11:50	12:50	Round Table Anna Jansen, Tanjala Gipson, Chris Kingswood, Carla Fladrowski, Inna Korob, Micaela Rozenberg Moderator John Bissler
12:50	13:00	Closing

13:00 | 14:00 Lunch









