



UNIVERSITÀ DEGLI STUDI DI TORINO

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# Novel insights into the relationship between KRIT1 and ROS homeostasis: KRIT1 loss-of-function causes a ROS-dependent upregulation of transcription factors involved in oxidative stress response.

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## 8<sup>th</sup> Annual Angioma Alliance CCM SCIENTIFIC MEETING

## November 15-16, 2012

DoubleTree Bethesda – Washington, DC Ballroom D

#### DAY 1 | THURSDAY, NOVEMBER 15<sup>TH</sup>

- 7:45 WELCOME & OPENING REMARKS Amy Akers & Sara Sukalich – Angioma Alliance
- SESSION I PROTEOMICS, STRUCTURE & FUNCTION DOUG MARCHUK, SESSION CHAIR
- 8:00 *New insights into the structure and function of CCM proteins* Titus Boggon – Yale University School of Medicine
- 8:20 **Proteomics characterization of CCM complexes** Anne-Claude Gingras – Samuel Lunenfeld Research Institute
- 8:40 Structural Basis of the Junctional Anchorage of the Cerebral Cavernous Malformations Complex Alexandre Gingras – University of California San Diego

#### SESSION II – SIGNALING

BRENT DERRY, SESSION CHAIR

- 9:00 **Defining the Ccm3 signaling pathway in a zebrafish model of CCM disease** Bilge Yoruk\*\* – Sick Kids Research Institute
- 9:20 *CCM3 and senescence* Juan Zalvide – University of Santiago de Compostela
- 9:40 COFFEE BREAK
- 10:00 **CCM3 functions in brain development** Angeliki Louvi – Yale School of Medicine
- 10:20 *CCM3 regulates endosome recycling in the C. elegans excretory cell* Ben Lant – Sick Kids Research Institute

10:40	Novel Endothelial Signaling in CCM
	Rebecca Stockton – University of California Los Angeles
11:00	Further Studies of Fasudil Treatment in Murine Models of Cerebral Cavernous Malformation Disease
	Robert Shenkar – University of Chicago
11:20	DISCUSSION OF SESSIONS I & II
12:00	Lunch   Oz Restaurant
SESSION	NIII – VASCULAR BIOLOGY & INFLAMMATION
	BRANT WEINSTEIN, SESSION CHAIR
1:00	The CCM2 paralogue CCM2L opposes canonical cerebral cavernous malformation signaling in endothelial cells during cardiovascular growth
	Xiangjian Zheng – University of Pennsylvania
1:20	Loss of Notch signaling in the adult endothelium: implications for CCM
	Andreas Fischer – German Cancer Research Center Heidelberg (DKFZ)
1:40	<b>The recombinant antibody construction and restricted B cell</b> <b>repertoire in Human Cerebral Cavernous Malformation (CCM)</b> Changbin Shi – University of Chicago
2:00	<b>Decreased KRIT1 expression leads to increased vascular</b> <b>permeability and modifies inflammatory responses in vivo.</b> Angela Glading – University of Rochester
2:20	<b>CCM2 intersects a novel pathway of cytokine mediated vascular instability</b> Dean Li – University of Utah
2:40	Coffee Break
Session	IV – LESION GENESIS

KEVIN WHITEHEAD, SESSION CHAIR

3:00 Novel insights into the relationship between KRIT1 and ROS homeostasis: KRIT1 loss-of-function causes a ROS-dependent upregulation of transcription factors involved in oxidative stress response

Saverio Francesco Retta – University of Torino

3:20	Exploring the Implications of a Two-Hit Mechanism in Cerebral Cavernous Malformations
	David McDonald** - Duke University Medical Center
3:40	<b>CCM3-dependent EC-SMC/pericyte interactions in CCM lesion</b> <b>development mouse models and mechanistic studies</b> Wang Min – Yale University
4:00	<b>Angiogenesis is Required for Cavernous Malformation Development</b> Kevin Whitehead – University of Utah
4:20	DISCUSSION OF SESSIONS III & IV
5:00	END OF DAY 1
7:00	DINNER   BALLROOM C

## DAY 2 | Friday, November 16th

8:30 WELCOME Connie Lee – Angioma Alliance & CCM3 Action

#### SESSION V – MAGNETIC RESONANCE IMAGING TECHNOLOGIES Leslie Morrison, Session Chair

- 8:40 Quantitative Iron Burden as a Biomarker of Cumulative Hemorrhages in Cerebral Cavernous Malformations: Studies in Mouse and Man Luying (Ryan) Li\*\* – West China Medical School of Sichuan University & University of Chicago
- 9:00 Novel Magnetic Resonance Imaging Biomarkers of Human CCM Disease: Dynamic Contrast-Enhanced Quantitative Perfusion Abdul Ghani Mikati<sup>\*\*</sup> – University of Chicago
- 9:20 White Matter Hyperintensities in CHM CCM1 Blaine Hart – University of New Mexico
- 9:40 COFFEE BREAK

#### SESSION VI – CLINICAL STUDIES

ISSAM AWAD, SESSION CHAIR

10:00 Spectrum of Human Causative Mutations in the KRIT1, CCM2 and PDCD10 Genes James Weber – PreventionGenetics

10:20	Clinical Factors Associated with Lesion Count in Familial Cerebral Cavernous Malformation Type 1 Patients with the Common Hispanic Mutation Hélène Choquet** – University of California San Francisco	
10:40	<b>Cutaneous Features of the CCM1-CHM Cohort</b> Leslie Morrison – University of New Mexico	
11:00	Outcome after surgical or conservative management of cerebral cavernous malformations: a prospective, population-based cohort study Margaret A. Horne <sup>**</sup> - University of Edinburgh	
11:20	DISCUSSION OF SESSION V & VI	
12:00	LUNCH   OZ RESTAURANT	
Session VII – Panel Discussion of Clinical Trails for CCM		
1:00	<b>Biomarkers</b> Issam Awad – University of Chicago	
1:10	<b>Recruitment Strategies</b> Leslie Morrison – University of New Mexico	
1:20	<b>Trials &amp; Research Consortia</b> William Young – University of California San Francisco	
1:30	<b>Food &amp; Drug Administration Perspective</b> Gumei Liu – Rare Diseases Program Office of New Drugs	
1:40	<b>National Institutes of Health Perspective</b> Claudia Moy – NINDS office of Clinical Research	
1:50	OPEN DISCUSSION	
3:00	CLOSE OF MEETING	

## MEETING SPONSORS

### ANGIOMA ALLIANCE NATIONAL INSTITUTE OF NEUROLOGICAL DISORDERS & STROKE CCM3 ACTION INNOLYST (PATIENT CROSSROADS)

\*\*Trainee Travel Award Winner