

Welcome
To the
9th Annual
Angioma Alliance

CCM SCIENTIFIC MEETING

WASHINGTON PLAZA HOTEL
WASHINGTON, DC

November 7-8, 2013



Angioma Alliance

Striving for Answers

MEETING AGENDA

Washington Plaza Hotel

Jefferson Room

Day 1 | Thursday, November 7th

8:45 Opening Remarks
Amy Akers, Angioma Alliance

SESSION I – CCM1 & CCM2 SIGNALING NETWORKS

Kelly Flemming, Session Chair

9:00 *The structure of the Ternary Complex of KRIT1 bound to both the Rap1 GTPase and the Heart of Glass (HEG1) cytoplasmic tail*
Alexandre Gingras – University of California San Diego

9:20 *Expanding the Heg-CCM Signaling Pathway Network*
John Mably – Boston Children's Hospital

9:40 *Exiting angiogenesis requires CCM proteins to attenuate proangiogenic Klf2 activity*
Salim Seyfried – Max Planck Institute of Molecular and Cellular Biology

10:00 Discussion

10:20 *Coffee Break*

SESSION II – IMMUNOLOGY & VASCULAR BIOLOGY

Doug Marchuk, Session Chair

10:30 *In situ humoral immunity in human cerebral cavernous malformations*
Changbin Shi – University of Chicago

10:50 *Reduced KRIT1 expression modifies vascular permeability by increasing oxidative stress*
Angela Glading – University of Rochester

11:10 *Role of KRIT1 in the pathogenesis of Cerebral Cavernous Malformations (CCM): From disease mechanisms toward pharmacological therapies*
Francesco Retta – University of Torino

11:30 Discussion

Noon *Lunch – Monroe Room*

SESSION III – MODEL SYSTEMS

Kevin Whitehead, Session Chair

1:00 *High-Content Screening Reveals CCM Pathophysiology and Potential Treatments*
Christopher Gibson – University of Utah

1:20 *Human endothelial progenitor-derived endothelial cells as a model system for Cerebral Cavernous Malformation disease*
Adriana Beltran, University of North Carolina at Chapel Hill

1:40 *A mouse model of CCM3*
Douglas Marchuk, Duke University Medical Center

2:00 Discussion

2:20 *Coffee Break*

SESSION IV – CCM3 MOLECULAR CHARACTERIZATION, SIGNALING & FUNCTION

Brent Derry, Session Chair

2:30 *Proteomic characterization of CCM3-GCKIII complexes*
James Knight, Lunenfeld-Tenanbaum Research Institute

2:50 *CCM3-Mst4-Cortactin complex regulates brain endothelial barrier function*
Anuska Andejejkovic-Zochowski – University of Michigan

3:10 *Using the Zebrafish Embryo to Decipher the CCM3 Signaling Pathway*
Ian Scott – The Hospital for Sick Children

3:30 *Coffee Break*

3:40 *CCM3/STRIPAK regulation of endosomal recycling and lumen maintenance in C. elegans seamless tubes*
Benjamin Lant – The Hospital for Sick Children

4:00 *Null mutations in Drosophila CCM3 and GCKIII cause tracheal tube dilations*
Amin Ghabrial – University of Pennsylvania School of Medicine

4:20 Discussion

4:55 *CCM3 Action Announcement*
Connie Lee – Angioma Alliance

5:00 END OF DAY 1 SESSIONS

6:30 Dinner at BUFFALO BILLIARDS, located on Dupont Circle

DAY 2 | Friday, November 8th

SESSION V – THE HUMAN CONDITION

Leslie Morrison, Session Chair

- 9:00 *Extensive Genetics of Cerebral Cavernous Malformations in Italy*
Silvana Penco, Italy
- 9:20 *Association of Common Variants in Immune Response Genes with Severity of Familial Cerebral Cavernous Malformations Type I*
Helene Choquet, UCSF
- 9:40 *Coffee Break*
- 9:50 *The Natural History of Incidentally Discovered Cavernous Malformations*
Kelly Flemming, The Mayo Clinic
- 10:10 *Lesion distribution and changes with age and radiation therapy in a population with familial CCM1*
Michael Golden, University of New Mexico
- 10:30 *Exceptional Aggressiveness of CCM3 Phenotype*
Issam Awad – University of Chicago
- 10:50 Discussion
- 11:20 *Lunch – Monroe Room*

SESSION VI – BIOMARKERS

Issam Awad, Session Chair

- 12:30 *Characterizing cerebral cavernous malformations with quantitative susceptibility mapping: further validation in humans and preliminary findings in mice*
Huan Tan – University of Chicago
- 12:50 *Updates in Permeability as a Biomarker of CCM Disease*
Abdul Ghani Mikati – University of Chicago
- 1:10 *Predictors of Functional outcome in Patients with CCM1 Common Hispanic Mutation*
Leslie Morrison – University of New Mexico
- 1:30 Discussion
- 1:50 *Coffee Break*

SESSION VII – CLINICAL TRIAL PREPAREDNESS

- 2:00 *Angioma Alliance Updates – Recruitment, Biobank & Clinical Site Development*
Amy Akers & Connie Lee – Angioma Alliance
- 3:30 Close of Meeting