



# **ANGIOMA** **ALLIANCE**

*Presents the 15<sup>th</sup> Annual*

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## **CCM SCIENTIFIC** **MEETING**

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**THE DOUBLETREE BY HILTON HOTEL**

**SILVER SPRING, MD**

**NOVEMBER 7-8, 2019**

## Day 1 | Thursday, November 7th, 2019

- 7:30 Registration, Pinnacle Grand Ballroom Foyer
- 7:30 Continental Breakfast, Connection Room
- 8:30 Welcome & Opening Remarks, Pinnacle Grand Ballroom

### **SESSION I – ANIMALS & PRECLINICAL STUDIES**

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Moderated by TBD

- 8:40 *Understanding the role of CCM3 in endothelial development and disease*  
Tvisha Misra • Sickkids Hospital
- 9:00 *Specific deletion of CCM3 in brain endothelium reliably models human cerebral cavernous malformations*  
Huanjiao Jenny Zhou • Yale
- 9:20 *Advancing CCM mouse models for pre-clinical therapeutic testing*  
Matthew Detter • Duke University
- 9:40 *Transcriptomes of Cerebral Cavernous Angiomas Clarify Mechanisms of Lesion Genesis and Maturation in Murine Pre-Clinical Models and Human Symptomatic Hemorrhage*  
Romuald Girard • University of Chicago
- 10:00 **DISCUSSION**
- 10:20 **BREAK**

### **SESSION II – NATURAL HISTORY & BIOMARKERS**

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Moderated by TBD

- 10:40 *Angioma Alliance updates - patient engagement, international collaborations & unraveling the genealogy of an American founder mutation*  
Connie Lee • Angioma Alliance
- 11:00 *Health-related Quality of Life in Cavernous Angioma Patients with Symptomatic Hemorrhage*  
Helen Kim • UCSF
- 11:20 *Subclinical Imaging Changes in Cerebral Cavernous Angiomas During Prospective Surveillance*  
Julian Carrion-Penagos • University of Chicago
- 11:40 *Common transcriptomic and biomarker signatures in the aging brain and in Mendelian neurovascular disease, cerebral cavernous malformation*  
Issam Awad • University of Chicago
- 12:00 **DISCUSSION**
- 12:20 **LUNCH - CONNECTION**

## SESSION III – HEMORRHAGE RISK & CLINICAL TRIALS

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Moderated by TBD

- 1:30 *Predictors of Intracranial hemorrhage in Familial Cerebral Cavernous Malformation Patients - BVMC Study Cohort*  
Atif Zafar • UNM
- 1:50 *Predictors of Initial Presentation with Hemorrhage in Patients with Cavernous Malformations - the role of clinical history and medications*  
Kelly Flemming • Mayo Clinic
- 2:10 *Association between statin or beta blocker drug use and hemorrhage from cerebral cavernous malformations*  
Susanna Zuurbier • Amsterdam University
- 2:30 *TREAT\_CCM - A multicenter randomized clinical trial on propranolol in familial cerebral cavernous malformations*  
Roberto Latini • Istituto Mario Negri
- 2:50 *Tempol clinical trials and developing a CCM-Health Index*  
Tracey Clayton • Recursion Pharmaceuticals
- 3:10 **DISCUSSION**
- 3:30 **GROUP PHOTO**

## POSTER SESSION | DISCOVERY ROOM (3:45-5 PM)

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*Symptomatic Brain Hemorrhages from Cavernous Angioma Following Botulinum Toxin Injections, and Suggested TLR/MEKK3 Mechanism*

Julian Carrion-Penagos • University of Chicago

*Characterizing Meningeal Lymphatic Development in Zebrafish*

Daniel Castranova • NIH

*The Cavernous Angioma Patient Registry – a tool for research & recruitment*

Kristen Dahlem • Angioma Alliance

*nfatc1 deficiency causes thoracic duct dilation during vascular development*

Alexandra Fister • NICHD/NIH

*Affected health domains in patients with brainstem cavernous malformations*

Kelly Flemming • Mayo Clinic

*Artery/Vein Plasticity After Vessel Injury*

Leah Greenspan • NIH

*Target sequencing for germline mutations in sporadic CCM patients*

Hiroki Hongo • University of Tokyo

*CCM1 and CCM3 cooperate to maintain intestinal function in C. elegans*

Sam Krempel • SickKids Hospital

*The role of CCM-3 in the ERK-5 pathway*  
Ben Lant • SickKids Hospital

*Selective ROCK Inhibitors Ameliorate CCM Lesions in an Acute Mouse Model*  
Matthew Lee • Cervello Therapeutics

*Endothelial cell clonal expansion in the development of Cerebral Cavernous Malformations*  
Matteo Malinverno • FIRC Institute

*Female Hormonal Therapy and Cavernous Angioma Hemorrhage*  
Jorge Marcondes • Universidade Federal Rio de Janeiro

*Variants in Inflammation-Related Genes Plus DNA Repair Enzymes and Aggressiveness in a CCM3 Brazilian Patient with Cerebral Cavernous Malformations.*  
Jorge Marcondes • Universidade Federal Rio de Janeiro

*KRIT1 deficiency promotes aortic endothelial dysfunction and atherosclerosis*  
Andrea Perrelli • University of Torino

*The role of MRCK-1 in biological tube development*  
Evelyn Popiel • The Hospital for Sick Children

*Adapting BioID for Use in Zebrafish to Investigate the Protein-Protein Interactions of CCM3*  
Shimon Rosenthal • University of Toronto

*A Brain Targeted Orally Available ROCK2 Inhibitor Benefits Mild and Aggressive Cavernous Angioma Disease*  
Robert Shenkar • University of Chicago

*Studying the origin and function of novel vascular-associated cells in the zebrafish meninges*  
Marina Venero Galanternik • NIH

*Characterizing Novel RHOA Mutant Alleles and their Effects on Vascular Integrity*  
Joseph Yano • NIH

*Prevalence of Obstructive Sleep Apnea (OSA) in Cerebral Cavernous Malformations*  
Atif Zafar • University of New Mexico

*Autoantigen(s) Trigger a Robust Immune Response in Cerebral Cavernous Malformations*  
Dondong Zhang & Abhinav Srinath • University of Chicago

5:00 **BREAK**

**WELCOME DINNER | MRS. K'S RESTAURANT & CELLAR (7-9 PM)**

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9201 Colesville Road, Silver Spring, MD

## Day 2 | Friday, November 8th, 2019

### **CONCURRENT SESSION SCIENTIFIC MEETING & FAMILY CONFERENCE**

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7:30 Continental Breakfast, Connection Room

8:30 Welcome & Introduction, Pinnacle Ballroom

8:40 **PLENARY PRESENTATION**

*Keynote Address to Patients, Families, and Investigators: Milestones and Our Road Ahead*

Issam Awad • University of Chicago

9:30 **BREAK**

### **SESSION IV – VASCULAR BIOLOGY & LESION DEVELOPMENT**

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Moderated by TBD

9:50 *Characterizing the function of RHOA in regulating vascular development and integrity in vivo*

Laura Pillay, NICHD/NIH

10:10 *Blood flow suppresses vascular anomalies in zebrafish model of cerebral cavernous malformations*

Claudia Rodel - Potsdam University

10:30 *Axonal Guidance Factors Regulate Invasion and Migration of Brain Endothelial Cells in Normal Development and Cerebrovascular Malformations*

Katie Fehnel - Boston Children's

10:50 **DISCUSSION**

11:45 **LUNCH – CONNECTION WITH FAMILY CONFERENCE ATTENDEES**

## SESSION V – VASCULAR DEVELOPMENT & LESION GENESIS

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Moderated by TBD

- 1:00 *CCM3, a protein mutated in cerebral cavernous malformations, is a signal transduction adapter*  
Kento Abe • University of Toronto
- 1:20 *Alternatively spliced isoforms reveal a novel type of PTB domain in CCM2 protein*  
Jun Zhang • Texas Tech University
- 1:40 *NgBR Regulates the Expression of CCM1/2 in Endothelial Cells via Histone Acetylation*  
Zhi Fang • New York University
- 2:00 *Ccm2l deletion aggravates cerebral cavernous malformation in Ccm2-deficient mice by activating MEKK3-KLF signaling pathway*  
Jaesung Choi • Centenary Institute
- 2:20 *Pharmacological inhibition of the HEG1-KRIT1 protein complex increases Kruppel-like Factors 4 and 2 expression in endothelial cells.*  
Miguel Lopez-Ramirez • UCSD
- 2:40 **DISCUSSION**
- 3:00 **BREAK WITH FAMILY CONFERENCE**
- 3:15 **CLOSE OF MEETING**

*Thank you to our sponsors!*



**RECURSION**  
p h a r m a c e u t i c a l s

**PREVENTION > GENETICS**

DISEASE PREVENTION THROUGH GENETIC TESTING