## 8<sup>th</sup> Annual Angioma Alliance

# **CCM SCIENTIFIC MEETING**

## November 15-16, 2012

 $\begin{array}{c} {\rm Double Tree\ Bethes da-Washington,\ DC} \\ {\it Ballroom\ D} \end{array}$ 

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DAV1 THURSDAY NOVEMBED 15TH

#### 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	<b>Proteomics characterization of CCM complexes</b> 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

Cavernous Malformation Disease Robert Shenkar – University of Chicago  11:20 DISCUSSION OF SESSIONS I & II  12:00 Lunch   Oz Restaurant  SESSION III — 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 The recombinant antibody construction and restricted B cell repertoire in Human Cerebral Cavernous Malformation (CCM) Changbin Shi — University of Chicago  2:00 Decreased KRIT1 expression leads to increased vascular permeability and modifies inflammatory responses in vivo. Angela Glading — University of Rochester  2:20 CCM2 intersects a novel pathway of cytokine mediated vascular instability	10:40	Novel Endothelial Signaling in CCM Rebecca Stockton – University of California Los Angeles
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#### Session IV – Lesion Genesis

2:40

COFFEE BREAK

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	CCM3-dependent EC-SMC/pericyte interactions in CCM lesion development mouse models and mechanistic studies Wang Min – Yale University
4:00	Angiogenesis is Required for Cavernous Malformation Development 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	e   Friday, November 16 <sup>th</sup>
8:30	<b>WELCOME</b> Connie Lee – Angioma Alliance & CCM3 Action
Session	N 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** – University of Chicago
9:20	White Matter Hyperintensities in CHM CCM1 Blaine Hart – University of New Mexico
9:40	Coffee Break
Session	N 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	Cutaneous Features of the CCM1-CHM Cohort 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** - University of Edinburgh
11:20	DISCUSSION OF SESSION V & VI
12:00	Lunch   Oz Restaurant
Session V	II – Panel Discussion of Clinical Trails for CCM
1:00	<b>Biomarkers</b> Issam Awad – University of Chicago
1:10	Recruitment Strategies

# William Young – University of California San Francisco 1:30 Food & Drug Administration Perspective Gumei Liu – Rare Diseases Program Office of New Drugs 1:40 National Institutes of Health Perspective

Trials & Research Consortia

Leslie Morrison – University of New Mexico

Claudia Moy – NINDS office of Clinical Research

## 1:50 OPEN DISCUSSION

1:20

3:00 CLOSE OF MEETING

### MEETING SPONSORS

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

\*\*Trainee Travel Award Winner