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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8th Annual Angioma Alliance

CCM Scientific Meeting

November 15-16, 2012

DoubleTree Bethesda – Washington, DC

Ballroom D

Day 1 | Thursday, November 15th

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 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  
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    **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 | 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** – 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 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 VII – Panel Discussion of Clinical Trails for CCM

1:00 Biomarkers
Issam Awad – University of Chicago

1:10 Recruitment Strategies
Leslie Morrison – University of New Mexico

1:20 Trials & Research Consortia
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
Claudia Moy – NINDS office of Clinical Research

1:50 Open Discussion

3:00 Close of Meeting

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