

Helen Kim - "Modifiers of disease severity in familial CCM"

Si segnala il seguente seminario:

Modifiers of disease severity

in familial Cerebral Cavernous Malformations

Martedì 24 giugno 2014 – ore 11.00

Aula Consiliare (Corpo F piano rialzato ^[1])

Università degli Studi di Brescia

Viale Europa, 11 - Brescia (mappa ^[2])

Relatrice:

Prof.ssa **Helen Kim**

Institute for Human Genetics (IHG ^[3])

Center for Cerebrovascular Research (CCR ^[4])

University of California, San Francisco (UCSF ^[5]), USA

Il seminario è aperto a tutti gli interessati.

Si allega la locandina ^[6] con preghiera di darne ampia diffusione

Organizzatori:

Prof. Marco M. Fontanella, Clinica Neurochirurgica, Università di Brescia

Prof. Alessandro Padovani, Clinica Neurologica, Università di Brescia

Prof. Saverio Francesco Retta, Dip. Scienze Cliniche e Biologiche, Università di Torino

per ulteriori informazioni:

Prof. Marco M. Fontanella, e-mail: marco.fontanella@unibs.it ^[7]

Helen Kim, MPH, PhD

Associate Professor of Anesthesia & Perioperative Care and of Epidemiology & Biostatistics, School of Medicine, University of California, San Francisco (UCSF) USA

Research interests:

Dr. Helen Kim focuses on identifying genetic factors that predispose individuals to stroke and outcomes after stroke. Her group studies families or individuals affected with cerebrovascular malformations, including arteriovenous malformations, cerebral cavernous malformations, intracranial aneurysms, and hereditary hemorrhagic telangiectasia. These cerebrovascular disorders, while distinct, share to some degree similar clinical symptoms and features. Current genetic epidemiology projects use whole genome SNP and expression arrays, and next-gen sequencing to identify genetic variation associated with disease susceptibility or progression. The long-term objective is to improve patient outcomes by identifying gene targets for therapy and factors to facilitate risk stratification of patients most amenable for clinical intervention. (see Helen Kim's full profile [8]).

Recent publications:

Choquet H, Nelson J, Pawlikowska L, McCulloch CE, Akers A, Baca B, Khan Y, Hart B, Morrison L, Kim H. **Association of cardiovascular risk factors with disease severity in cerebral cavernous malformation type 1 subjects with the common Hispanic mutation.** Cerebrovasc Dis. 2014; 37(1):57-63. ?View in: PubMed [9]

Bendjilali N, Kim H, Weinsheimer S, Guo DE, Kwok PY, Zaroff JG, Sidney S, Lawton MT, McCulloch CE, Koeleman BP, Klijn CJ, Young WL, Pawlikowska L. **A genome-wide investigation of copy number variation in patients with sporadic brain arteriovenous malformation.** PLoS One. 2013; 8(10):e71434. ?View in: PubMed [10]

Zaroff JG, Leong J, Kim H, Young WL, Cullen SP, Rao VA, Sorel M, Quesenberry CP, Sidney S. **Cardiovascular predictors of long-term outcomes after non-traumatic subarachnoid hemorrhage.** Neurocrit Care. 2012 Dec; 17(3):374-81. View in: PubMed [11]

Rodríguez-Hernández A, Kim H, Pourmohamad T, Young WL, Lawton MT. **Cerebellar arteriovenous malformations: anatomic subtypes, surgical results, and increased predictive accuracy of the supplementary grading system.** Neurosurgery. 2012 Dec; 71(6):1111-24. View in: PubMed [12]

Boshuisen K, Brundel M, de Kovel CG, Letteboer TG, Rinkel GJ, Westermann CJ, Kim H, Pawlikowska L, Koeleman BP, Klijn CJ. **Polymorphisms in ACVRL1 and Endoglin Genes are Not Associated with Sporadic and HHT-Related Brain AVMs in Dutch Patients.** Transl Stroke Res. 2013 Jun; 4(3):375-8. View in: PubMed [13]

Khush KK, Pawlikowska L, Menza RL, Goldstein BA, Hayden V, Nguyen J, Kim H, Poon A, Sapru A, Matthay MA, Kwok PY, Young WL, Baxter-Lowe LA, Zaroff JG. **Beta-adrenergic receptor polymorphisms and cardiac graft function in potential organ donors.** Am J Transplant. 2012 Dec; 12(12):3377-86. ?View in: PubMed [14]

Nishida T, Faughnan ME, Krings T, Chakinala M, Gossage JR, Young WL, Kim H, Pourmohamad T, Henderson KJ, Schrum SD, James M, Quinnine N, Bharatha A, Terbrugge KG, White RI. **Brain arteriovenous malformations associated with hereditary hemorrhagic telangiectasia: gene-phenotype correlations.**

Am J Med Genet A. 2012 Nov; 158A(11):2829-34. ?View in: PubMed [15]

Weinsheimer S, Brettman AD, Pawlikowska L, Wu DC, Mancuso MR, Kuhnert F, Lawton MT, Sidney S, Zaroff JG, McCulloch CE, Young WL, Kuo C, Kim H. **G Protein-Coupled Receptor 124 (GPR124) Gene Polymorphisms and Risk of Brain Arteriovenous Malformation.** Transl Stroke Res. 2012 Dec; 3(4):418-27. ?View in: PubMed [16]

Kim H, Pourmohamad T, Westbroek EM, McCulloch CE, Lawton MT, Young WL. **Evaluating performance of the Spetzler-Martin supplemented model in selecting patients with brain arteriovenous malformation for surgery.** Stroke. 2012 Sep; 43(9):2497-9. ?View in: PubMed [17]

Kahle MP, Lee B, Pourmohamad T, Cunningham A, Su H, Kim H, Chen Y, McCulloch CE, Barbaro NM, Lawton MT, Young WL, Bix GJ. **Perlecan domain V is upregulated in human brain arteriovenous malformation and could mediate the vascular endothelial growth factor effect in lesional tissue.** Neuroreport. 2012 Jul 11; 23(10):627-30. ?View in: PubMed [18]

Westbroek EM, Pawlikowska L, Lawton MT, McCulloch CE, Young WL, Kim H. **Brain-derived neurotrophic factor Val66Met polymorphism predicts worse functional outcome after surgery in patients with unruptured brain arteriovenous malformation.** Stroke. 2012 Aug; 43(8):2255-7. ?View in: PubMed [19]

Lingua Italiano

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