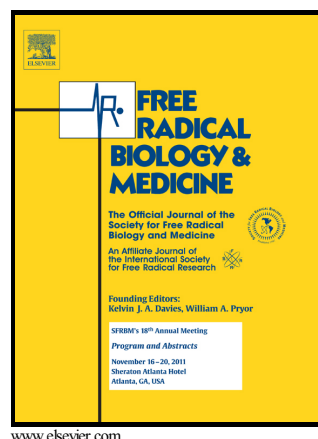


Author's Accepted Manuscript

Cytochrome P450 and Matrix Metalloproteinase
Genetic Modifiers of Disease Severity in Cerebral
Cavernous Malformation type 1

Hélène Choquet, Eliana Trapani, Luca Goitre,
Lorenza Trabalzini, Amy Akers, Marco Fontanella,
Blaine L. Hart, Leslie A. Morrison, Ludmila
Pawlikowska, Helen Kim, Saverio Francesco Retta



PII: S0891-5849(16)00009-5
DOI: <http://dx.doi.org/10.1016/j.freeradbiomed.2016.01.008>
Reference: FRB12719

To appear in: *Free Radical Biology and Medicine*

Received date: 12 November 2015

Accepted date: 13 January 2016

Cite this article as: Hélène Choquet, Eliana Trapani, Luca Goitre, Lorenza Trabalzini, Amy Akers, Marco Fontanella, Blaine L. Hart, Leslie A. Morrison, Ludmila Pawlikowska, Helen Kim and Saverio Francesco Retta, Cytochrome P450 and Matrix Metalloproteinase Genetic Modifiers of Disease Severity in Cerebral Cavernous Malformation type 1, *Free Radical Biology and Medicine*, <http://dx.doi.org/10.1016/j.freeradbiomed.2016.01.008>

This is a PDF file of an unedited manuscript that has been accepted for publication. As a service to our customers we are providing this early version of the manuscript. The manuscript will undergo copyediting, typesetting, and review of the resulting galley proof before it is published in its final citable form. Please note that during the production process errors may be discovered which could affect the content, and all legal disclaimers that apply to the journal pertain.

Cytochrome P450 and Matrix Metalloproteinase Genetic Modifiers of Disease Severity in Cerebral Cavernous Malformation type 1

Hélène Choquet¹, Eliana Trapani^{2,11}, Luca Goitre^{2,11}, Lorenza Trabalzini^{3,11}, Amy Akers⁴, Marco Fontanella^{5,11}, Blaine L. Hart⁶, Leslie A. Morrison^{7,8}, Ludmila Pawlikowska^{1,9}, Helen Kim^{1,9,10}, Saverio Francesco Retta^{2,11§}

¹Center for Cerebrovascular Research, Department of Anesthesia and Perioperative Care, University of California, San Francisco, California, USA

²Department of Clinical and Biological Sciences, University of Torino, Orbassano (TO), Italy

³Department of Biotechnology, Chemistry and Pharmacy, University of Siena, Siena, Italy

⁴Angioma Alliance, Durham, North Carolina, USA

⁵Department of Neurosurgery, Spedali Civili and University of Brescia, Brescia, Italy

⁶Departments of Radiology (B.L.H.), University of New Mexico, Albuquerque, New Mexico, USA

⁷Neurology University of New Mexico, Albuquerque, New Mexico, USA

⁸Pediatrics (L.A.M.), University of New Mexico, Albuquerque, New Mexico, USA

⁹Institute for Human Genetics, University of California, San Francisco, California, USA

¹⁰Department of Epidemiology and Biostatistics, University of California, San Francisco, California, USA

¹¹CCM Italia research network (www.ccmitalia.unito.it)

[§]Address Correspondence to: Prof. Saverio Francesco Retta, Department of Clinical and Biological Sciences, University of Torino, Italy, Regione Gonzole, 10 - 10043 Orbassano (Torino), Tel.: (39) 011-6706426. Fax: (39) 011-9038639, E-mail: francesco.retta@unito.it

Abstract

Background: Familial Cerebral Cavernous Malformation type 1 (CCM1) is an autosomal dominant disease caused by mutations in the Krev Interaction Trapped 1 (*KRIT1/CCM1*), and characterized by multiple brain lesions. CCM lesions manifest across a range of different phenotypes, including wide differences in lesion number, size and susceptibility to intracerebral hemorrhage (ICH). Oxidative stress plays an important role in cerebrovascular disease pathogenesis, raising the possibility that inter-individual variability in genes related to oxidative stress may contribute to the phenotypic differences observed in CCM1 disease. Here, we investigated whether candidate oxidative stress-related cytochrome P450 (CYP) and matrix metalloproteinase (MMP) genetic markers grouped by superfamilies, families or genes, or analyzed individually influence the severity of CCM1 disease.

Download English Version:

<https://daneshyari.com/en/article/8268105>

Download Persian Version:

<https://daneshyari.com/article/8268105>

[Daneshyari.com](https://daneshyari.com)