CCM2

CCM2 binds to MEKK3.

Mutations in the genes KRIT1, CCM2, and PDCD10 are known to result in the formation of cerebral cavernous malformations (CCMs).

Although these genes have been known to be associated with CCMs since the 1990s, numerous discoveries have been made that better elucidate how they and their subsequent protein products are involved in CCM pathogenesis. Since our last review of the molecular genetics of CCM pathogenesis in 2012, breakthroughs include a more thorough understanding of the protein structures of the gene products, involvement with integrin proteins, and MEKK3 signaling pathways, and the importance of CCM2-PDCD10 interactions [1)].

The CCM2 gene contains 10 coding exons and an alternatively spliced exon 1B. This gene is located on chromosome 7p13 and loss of function mutations on CCM2 lead to the onset of Cerebral Cavernous Malformations (CCM) illness.

Malcavernin is a protein that in humans is encoded by the CCM2 gene.

The normal function of malcavernin is to act as a scaffold for a variety of signaling complexes including p38 MAP Kinase.

This protein is also involved in regulating the cellular localization of the KRIT1 protein and acts with the Rho Kinase signaling pathway to maintain normal blood vessel structure.

1)
Baranoski JF, Kalani MY, Przybylowski CJ, Zabramski JM. Cerebral Cavernous Malformations: Review of the Genetic and Protein-Protein Interactions Resulting in Disease Pathogenesis. Front Surg. 2016 Nov 14;3:60. Review. PubMed PMID: 27896269.