Krev interaction trapped protein 1 is a protein that in humans is encoded by the CCM1 gene.

This gene contains 16 coding exons and is located on chromosome 7q21.2. Loss of function mutations in CCM1 result in the onset of Cerebral cavernous malformation.

Cerebral cavernous malformations (CCMs) are vascular malformations in the brain and spinal cord made of dilated capillary vessels.

The normal CCM1 protein, KRIT1 (Krev Interaction Trapped), is 736 amino acids in length and has a variety of functions. KRIT1 has been shown to interact with multiple signaling pathways including; ITGB1BP1., reactive oxygen species, cell death, and angiogenesis.

Related to the CCM illness, this protein is required for maintaining the structural integrity of the vasculature.

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. ¹⁾.

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.

From: https://neurosurgerywiki.com/wiki/ - **Neurosurgery Wiki**

Permanent link: https://neurosurgerywiki.com/wiki/doku.php?id=krit1

Last update: 2024/06/07 02:55



1/1