



## Malcavernin (H-85): sc-68412

### BACKGROUND

Cerebral cavernous malformation (CCM) is an autosomal dominant or sporadic neurovascular disease marked by vascular anomalies located mostly in the central nervous system that can cause stroke, seizures, cerebral hemorrhages, headaches and focal neurologic deficits. CCM is caused by mutations in one of three genes: CCM1, CCM2 or CCM3. CCM1 encodes the protein KRIT1, CCM2 encodes the protein Malcavernin and CCM3 shares its name with the protein it encodes. Malcavernin, also designated cerebral cavernous malformations 2 protein, is a scaffolding protein for MEK kinase-3. Like KRIT1, Malcavernin is expressed in a variety of human organs, including the arterial vascular endothelium, pyramidal neurons, astrocytes and their foot processes. In addition, Malcavernin is expressed in various epithelial cells that are required for the formation of the blood-organ barrier. Malcavernin is localized to the cytoplasm but is known to shuttle to and from the nucleus. Due to its lack of a nuclear export signal or nuclear localization signal, it is believed that Malcavernin accomplishes this shuttling via an attachment to KRIT1, which contains a nuclear localization signal. Two isoforms exist for Malcavernin. Isoform 1 represents the full length protein while isoform 2 contains an alternative four amino acid sequence rather than the first ten residues of isoform 1.

### REFERENCES

1. Lawton, M.T., et al. 2005. Giant infiltrative cavernous malformation: clinical presentation, intervention, and genetic analysis: case report. *Neurosurgery* 55: 979-980.
2. Guclu, B., et al. 2005. Mutations in apoptosis-related gene, PDCD10, cause cerebral cavernous malformation 3. *Neurosurgery* 57: 1008-1013.
3. Guclu, B., et al. 2005. Cerebral venous malformations have distinct genetic origin from cerebral cavernous malformations. *Stroke* 36: 2479-2480.
4. Zawistowski, J.S., et al. 2005. CCM1 and CCM2 protein interactions in cell signaling: implications for cerebral cavernous malformations pathogenesis. *Hum. Mol. Genet.* 14: 2521-2531.
5. Seker, A., et al. 2006. CCM2 expression parallels that of CCM1. *Stroke* 37: 518-523.
6. Labauge, P., et al. 2007. Genetics of cavernous angiomas. *Lancet Neurol.* 6: 237-244.
7. Ortiz, L., et al. 2007. Study of cerebral cavernous malformation in Spain and Portugal: high prevalence of a 14 bp deletion in exon 5 of MGC4607 (CCM2 gene). *J. Neurol.* 254: 322-326.
8. Zhang, J., et al. 2007. Interaction between KRIT1 and Malcavernin: implications for the pathogenesis of cerebral cavernous malformations. *Neurosurgery* 60: 353-359.

### CHROMOSOMAL LOCATION

Genetic locus: CCM2 (human) mapping to 7p13; Ccm2 (mouse) mapping to 11 A1.

### RESEARCH USE

For research use only, not for use in diagnostic procedures.

### SOURCE

Malcavernin (H-85) is a rabbit polyclonal antibody raised against amino acids 216-300 mapping within an internal region of Malcavernin of human origin.

### PRODUCT

Each vial contains 200 µg IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

### APPLICATIONS

Malcavernin (H-85) is recommended for detection of Malcavernin of mouse and human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunoprecipitation [1–2 µg per 100–500 µg of total protein (1 ml of cell lysate)], immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

Suitable for use as control antibody for Malcavernin siRNA (h): sc-62594 and Malcavernin siRNA (m): sc-62595.

Molecular Weight of Malcavernin: 49 kDa.

### RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use goat anti-rabbit IgG-HRP: sc-2004 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible goat anti-rabbit IgG-HRP: sc-2030 (dilution range: 1:2000-1:5000), Cruz Marker™ Molecular Weight Standards: sc-2035, TBS Blotto A Blocking Reagent: sc-2333 and Western Blotting Luminol Reagent: sc-2048. 2) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml). 3) Immunofluorescence: use goat anti-rabbit IgG-FITC: sc-2012 (dilution range: 1:100-1:400) or goat anti-rabbit IgG-TR: sc-2780 (dilution range: 1:100-1:400) with UltraCruz™ Mounting Medium: sc-24941.

### STORAGE

Store at 4° C, \*\*DO NOT FREEZE\*\*. Stable for one year from the date of shipment. Non-hazardous. No MSDS required.