



Malcavernin (h): 293T Lysate: sc-116673

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.

CHROMOSOMAL LOCATION

Genetic locus: CCM2 (human) mapping to 7p13.

PRODUCT

Malcavernin (h): 293T Lysate represents a lysate of human Malcavernin transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

STORAGE

Store at -20° C. Repeated freezing and thawing should be minimized. Sample vial should be boiled once prior to use. Non-hazardous. No MSDS required.

APPLICATIONS

Malcavernin (h): 293T Lysate is suitable as a Western Blotting positive control for human reactive Malcavernin antibodies. Recommended use: 10-20 µl per lane.

Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

RESEARCH USE

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

PROTOCOLS

See our web site at www.scbt.com for detailed protocols and support products.