

hamartin (18): sc-136436

BACKGROUND

Tuberous sclerosis complex (TSC) is an autosomal dominant genetic disorder characterized by mental retardation and the widespread development of distinctive tumors termed hamartomas. Two different genetic loci have been linked to TSC; one of these loci, the tuberous sclerosis-2 gene (TSC2), encodes a protein called tuberin and the other loci, tuberous sclerosis-1 gene (TSC1), encodes a protein called hamartin. Tuberin and hamartin interact with each other forming a cytoplasmic complex. Hamartin interacts with the ezrin-radixin-moesin (ERM) family of Actin-binding proteins and inhibition of hamartin activity results in loss of cell adhesion. Hamartin is present in most adult tissues with strong expression in brain, heart, and kidney.

REFERENCES

- Young, J., et al. 1998. The genetic basis of tuberous sclerosis. *Mol. Med. Today* 4: 313-319.
- Plank, T.L., et al. 1998. Hamartin, the product of the tuberous sclerosis 1 (TSC1) gene, interacts with tuberin and appears to be localized to cytoplasmic vesicles. *Cancer Res.* 58: 4766-4770.
- The European Chromosome 16 Tuberous Sclerosis Consortium. 1993. Identification and characterization of the tuberous sclerosis gene on chromosome 16. *Cell* 75: 1305-1315.
- van Slegtenhorst, M., et al. 1997. Identification of the tuberous sclerosis gene TSC1 on chromosome 9q34. *Science* 277: 805-808.
- Nellist, M., et al. 1999. Characterization of the cytosolic tuberin-hamartin complex. Tuberin is a cytosolic chaperone for hamartin. *J. Biol. Chem.* 274: 35647-35652.
- Plank, T.L., et al. 1999. The expression of hamartin, the product of the TSC1 gene, in normal human tissues and in TSC1- and TSC2-linked angiomyolipomas. *Mod. Pathol.* 12: 539-545.
- Fukuhara, S., et al. 2000. A new twist for the tumour suppressor hamartin. *Nat. Cell Biol.* 2: E76-E78.
- Lamb, R.F., et al. 2000. The TSC1 tumour suppressor hamartin regulates cell adhesion through ERM proteins and the GTPase Rho. *Nat. Cell Biol.* 2: 281-287.

CHROMOSOMAL LOCATION

Genetic locus: TSC1 (human) mapping to 9q34.13; Tsc1 (mouse) mapping to 2 A3.

SOURCE

hamartin (18) is a mouse monoclonal antibody raised against amino acids 932-1046 of hamartin of human origin.

PRODUCT

Each vial contains 200 µg IgG₁ kappa light chain in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

APPLICATIONS

hamartin (18) is recommended for detection of hamartin of mouse, rat and human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000) and immunoprecipitation [1-2 µg per 100-500 µg of total protein (1 ml of cell lysate)].

hamartin (18) is also recommended for detection of hamartin in additional species, including canine.

Suitable for use as control antibody for hamartin siRNA (h): sc-37437, hamartin siRNA (m): sc-37438, hamartin siRNA (r): sc-270022, hamartin shRNA Plasmid (h): sc-37437-SH, hamartin shRNA Plasmid (m): sc-37438-SH, hamartin shRNA Plasmid (r): sc-270022-SH, hamartin shRNA (h) Lentiviral Particles: sc-37437-V, hamartin shRNA (m) Lentiviral Particles: sc-37438-V and hamartin shRNA (r) Lentiviral Particles: sc-270022-V.

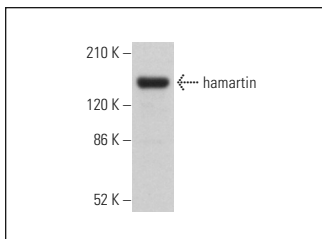
Molecular Weight of hamartin: 130 kDa.

Positive Controls: SJRH30 cell lysate: sc-2287 or WI-38 whole cell lysate: sc-364260.

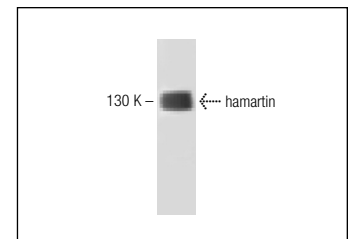
RECOMMENDED SUPPORT REAGENTS

To ensure optimal results, the following support reagents are recommended: 1) Western Blotting: use m-IgGκ BPHRP: sc-516102 or m-IgGκ BPHRP (Cruz Marker): sc-516102-CM (dilution range: 1:1000-1:10000), Cruz Marker™ Molecular Weight Standards: sc-2035, UltraCruz® Blocking Reagent: sc-516214 and Western Blotting Luminol Reagent: sc-2048. 2) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml).

DATA



hamartin (18): sc-136436. Western blot analysis of hamartin expression in SJRH30 whole cell lysate.



hamartin (18): sc-136436. Western blot analysis of hamartin expression in WI-38 whole cell lysate.

SELECT PRODUCT CITATIONS

- Dos Santos Junior, E.F., et al. 2018. Calcitriol reverses the down-regulation pattern of tuberous sclerosis complex genes in an *in vitro* calcification model. *J. Mol. Neurosci.* 64: 140-143.

STORAGE

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

RESEARCH USE

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