

# hamartin (E-20): sc-12080

## 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

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2. van Slegtenhorst, M., et al. 1997. Identification of the tuberous sclerosis gene TSC1 on chromosome 9q34. *Science* 277: 805-808.
3. Young, J., et al. 1998. The genetic basis of tuberous sclerosis. *Mol. Med. Today* 4: 313-319.
4. 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.
5. 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.
6. 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.
7. Fukuhara, S., et al. 2000. A new twist for the tumour suppressor hamartin. *Nat. Cell Biol.* 2: E76-E78.
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## CHROMOSOMAL LOCATION

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

## SOURCE

hamartin (E-20) is an affinity purified goat polyclonal antibody raised against a peptide mapping near the N-terminus of hamartin 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.

Blocking peptide available for competition studies, sc-12080 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

## APPLICATIONS

hamartin (E-20) 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), immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500), immunohistochemistry (including paraffin-embedded sections) (starting dilution 1:50, dilution range 1:50-1:500) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

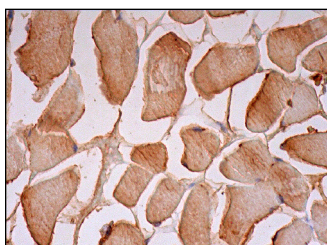
hamartin (E-20) is also recommended for detection of hamartin in additional species, including equine, canine and bovine.

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: MDA-MB-231 cell lysate: sc-2232 or MDA-MB-468 cell lysate: sc-2282.

## DATA



hamartin (E-20): sc-12080. Immunoperoxidase staining of formalin fixed, paraffin-embedded human skeletal muscle tissue showing membrane and cytoplasmic staining of myocytes.

## 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.

## PROTOCOLS

See our web site at [www.scbt.com](http://www.scbt.com) or our catalog for detailed protocols and support products.

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