SANTA CRUZ BIOTECHNOLOGY, INC.

hamartin (1-300): sc-4325 WB

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 180 kDa protein called tuberin. The other loci, tuberous sclerosis-1 gene (TSC1), encodes a 130 kDa protein called hamartin. Tuberin and hamartin interact with each other forming a cystoplasmic 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

1. The European Chromosome 16 Tuberous Sclerosis Consortium. 1993 Identification and characterization of the tuberous sclerosis gene on chromosome 16. Cell 75: 1305-1315.
2. van Slegtenhorst, M., de Hoogt, R., Hermans, C., Nellist, M., Janssen, B., Verhoef, S., Lindhout, D., et al. 1997. Identification of the tuberous sclerosis gene TSC1 on chromosome 9q34. Science 277: 805-808.
3. Young, J. and Povey, S. 1998. The genetic basis of tuberous sclerosis. Mol. Med. Today 4: 313-319.
4. Plank, T.L., Yeung, R.S., and Henske, E.P. 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., van Slegtenhorst, M.A., Goedbloed, M., van den Ouweland, A.M., Halley, D.J., and van der Sluijs, P. 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., Logginidou, H., Klein-Szanto, A., and Henske, E.P. 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. and Gutkind, J.S. 2000. A new twist for the tumour suppressor hamartin. Nat. Cell Biol. 2: E76-E78.
8. Lamb, R.F., Roy, C., Diefenbach, T.J., Vinters, H.V., Johnson, M.W., Jay, D.G., and Hall, A. 2000. The TSC1 tumour suppressor hamartin regulates cell adhesion through ERM proteins and the GTPase Rho. Nat. Cell Biol. 2: 281-287.

SOURCE

hamartin (1-300) is expressed in *E. coli* as a 60 kDa tagged fusion protein corresponding to amino acids 1-300 of hamartin of human origin.

PRODUCT

hamartin (1-300) is purified from bacterial lysates (>98%) by column chromatography; supplied as 10 µg in 0.1 ml SDS-PAGE loading buffer.

APPLICATIONS

hamartin (1-300) is suitable as a Western blotting control for sc-12080 and sc-13013.

STORAGE

Store at -20° C; stable for one year from the date of shipment.

RESEARCH USE

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