

folliculin (h2): 293T Lysate: sc-112237

BACKGROUND

Birt-Hogg-Dubé (BHD) syndrome is a rare autosomal dominant cancer syndrome characterized by kidney tumors, benign tumors of the hair follicle and spontaneous pneumothorax. BHD is also associated with neoplastic colonic polyps. The BHD gene maps to chromosome 17p11.2 and encodes the protein folliculin. Folliculin is widely expressed. Notably, folliculin is expressed in the kidney, lung and skin where BHD tumors arise. Specifically, the (C)8 tract in exon 11 is a mutational hot spot in BHD. BHD appears to have reduced penetrance or late onset. In a study of the renal tumors in 30 BHD patients, preoperative computed tomography scans detect a mean of 5.3 tumors per patient with a range 1-28 tumors. Multiple and bilateral tumors appear at a mean of 50.7 years.

REFERENCES

1. Balus, L., Fazio, M., Sacerdoti, G., Morrone, A. and Marmo, W. 1983. Fibrofolliculoma, trichodiscoma and acrochordon. The Birt-Hogg-Dubé syndrome. *Ann. Dermatol. Venereol.* 110: 601-609.
2. Schmidt, L.S., Warren, M.B., Nickerson, M.L., Weirich, G., Matrosova, V., Toro, J.R., Turner, M.L., Duray, P., Merino, M., Hewitt, S., Pavlovich, C.P., Glenn, G., Greenberg, C.R., Linehan, W.M. and Zbar, B. 2001. Birt-Hogg-Dubé syndrome, a genodermatosis associated with spontaneous pneumo-thorax and kidney neoplasia, maps to chromosome 17p11.2. *Am. J. Hum. Genet.* 69: 876-882.
3. Khoo, S.K., Giraud, S., Kahnoski, K., Chen, J., Motorna, O., Nickolov, R., Binet, O., Lambert, D., Friedel, J., Lévy, R., Ferlicot, S., Wolkenstein, P., Hammel, P., Bergerheim, U., Hedblad, M.A., Bradley, M., Teh, B.T., Nordenskjöld, M. and Richard, S. 2002. Clinical and genetic studies of Birt-Hogg-Dubé syndrome. *J. Med. Genet.* 39: 906-912.
4. Nickerson, M.L., Warren, M.B., Toro, J.R., Matrosova, V., Glenn, G., Turner, M.L., Duray, P., Merino, M., Choyke, P., Pavlovich, C.P., Sharma, N., Walther, M., Munroe, D., Hill, R., Maher, E., Greenberg, C., Lerman, M.I., Linehan, W.M., Zbar, B. and Schmidt, L.S. 2002. Mutations in a novel gene lead to kidney tumors, lung wall defects, and benign tumors of the hair follicle in patients with the Birt-Hogg-Dubé syndrome. *Cancer Cell* 2: 157-164.
5. Pavlovich, C.P., Walther, M.M., Eyler, R.A., Hewitt, S.M., Zbar, B., Linehan, W.M. and Merino, M.J. 2002. Renal tumors in the Birt-Hogg-Dubé syndrome. *Am. J. Surg. Pathol.* 26: 1542-1552.
6. Warren, M.B., Torres-Cabala, C.A., Turner, M.L., Merino, M.J., Matrosova, V.Y., Nickerson, M.L., Ma, W., Linehan, W.M., Zbar, B. and Schmidt, L.S. 2004. Expression of Birt-Hogg-Dubé gene mRNA in normal and neoplastic human tissues. *Mod. Pathol.* 17: 998-1011.
7. Nagy, A., Zoubakov, D., Stupar, Z. and Kovacs, G. 2004. Lack of mutation of the folliculin gene in sporadic chromophobe renal cell carcinoma and renal oncocytoma. *Int. J. Cancer* 109: 472-475.

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.

CHROMOSOMAL LOCATION

Genetic locus: FLCN (human) mapping to 17p11.2.

PRODUCT

folliculin (h2): 293T Lysate represents a lysate of human folliculin transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

APPLICATIONS

folliculin (h2): 293T Lysate is suitable as a Western Blotting positive control for human reactive folliculin 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.

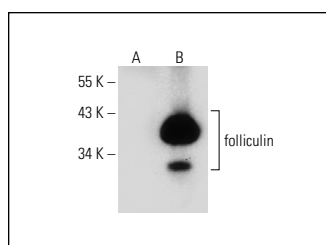
folliculin (D-4): sc-271558 is recommended as a positive control antibody for Western Blot analysis of enhanced human folliculin expression in folliculin transfected 293T cells (starting dilution 1:100, dilution range 1:100-1:1,000).

RECOMMENDED SUPPORT REAGENTS

To ensure optimal results, the following support reagents are recommended:

1) Western Blotting: use m-IgGκ BP-HRP: sc-516102 or m-IgGκ BP-HRP (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.

DATA



folliculin (D-4): sc-271558. Western blot analysis of folliculin expression in non-transfected: sc-117752 (A) and human folliculin transfected: sc-112237 (B) 293T whole cell lysates.

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.