

# folliculin (D-4): sc-271558

## BACKGROUND

Birt-Hogg-Dube (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

- Balus, L., et al. 1983. Fibrofolliculoma, trichodiscoma and acrochordon. The Birt-Hogg-Dube syndrome. *Ann. Dermatol. Venereol.* 110: 601-609.
- Schmidt, L.S., et al. 2001. Birt-Hogg-Dube syndrome, a genodermatosis associated with spontaneous pneumothorax and kidney neoplasia, maps to chromosome 17p11.2. *Am. J. Hum. Genet.* 69: 876-882.
- Khoo, S.K., et al. 2002. Clinical and genetic studies of Birt-Hogg-Dube syndrome. *J. Med. Genet.* 39: 906-912.

## CHROMOSOMAL LOCATION

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

## SOURCE

folliculin (D-4) is a mouse monoclonal antibody specific for an epitope mapping between amino acids 555-579 at the C-terminus of folliculin of human origin.

## PRODUCT

Each vial contains 200 µg IgG<sub>1</sub> kappa light chain in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

folliculin (D-4) is available conjugated to agarose (sc-271558 AC), 500 µg/0.25 ml agarose in 1 ml, for IP; to HRP (sc-271558 HRP), 200 µg/ml, for WB, IHC(P) and ELISA; to either phycoerythrin (sc-271558 PE), fluorescein (sc-271558 FITC), Alexa Fluor<sup>®</sup> 488 (sc-271558 AF488), Alexa Fluor<sup>®</sup> 546 (sc-271558 AF546), Alexa Fluor<sup>®</sup> 594 (sc-271558 AF594) or Alexa Fluor<sup>®</sup> 647 (sc-271558 AF647), 200 µg/ml, for WB (RGB), IF, IHC(P) and FCM; and to either Alexa Fluor<sup>®</sup> 680 (sc-271558 AF680) or Alexa Fluor<sup>®</sup> 790 (sc-271558 AF790), 200 µg/ml, for Near-Infrared (NIR) WB, IF and FCM.

Blocking peptide available for competition studies, sc-271558 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% stabilizer protein).

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## STORAGE

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

## APPLICATIONS

folliculin (D-4) is recommended for detection of folliculin of human origin by Western Blotting (starting dilution 1:100, dilution range 1:100-1:1000), immunoprecipitation [1-2 µg per 100-500 µg of total protein (1 ml of cell lysate)], immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

Suitable for use as control antibody for folliculin siRNA (h): sc-37412, folliculin shRNA Plasmid (h): sc-37412-SH and folliculin shRNA (h) Lentiviral Particles: sc-37412-V.

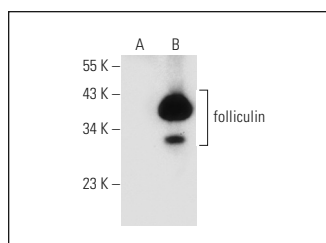
Molecular Weight of folliculin: 66 kDa.

Positive Controls: Caki-1 cell lysate: sc-2224 or folliculin (h): 293 Lysate: sc-112237.

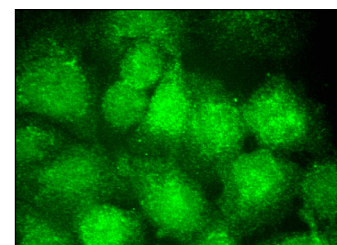
## RECOMMENDED SUPPORT REAGENTS

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<sup>™</sup> Molecular Weight Standards: sc-2035, UltraCruz<sup>®</sup> 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). 3) Immunofluorescence: use m-IgGκ BP-FITC: sc-516140 or m-IgGκ BP-PE: sc-516141 (dilution range: 1:50-1:200) with UltraCruz<sup>®</sup> Mounting Medium: sc-24941 or UltraCruz<sup>®</sup> Hard-set Mounting Medium: sc-359850.

## DATA



folliculin (D-4): sc-271558. Western blot analysis of folliculin expression in non-transfected: sc-110760 (A) and human folliculin transfected: sc-112237 (B) 293 whole cell lysates.



folliculin (D-4): sc-271558. Immunofluorescence staining of formalin-fixed A-431 cells showing nuclear and cytoplasmic localization.

## SELECT PRODUCT CITATIONS

- Wu, X., et al. 2016. FLCN maintains the leucine level in lysosome to stimulate mTORC1. *PLoS ONE* 11: e0157100.
- Nada, S. and Okada, M. 2020. Genetic dissection of Ragulator structure and function in amino acid-dependent regulation of mTORC1. *J. Biochem.* 168: 621-632.

## RESEARCH USE

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