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Rabbit Anti-FLCN antibody

SL6007R

Product Name:	FLCN
Chinese Name:	卵巢滤泡激素抗体(BHD综合征)
Alias:	BHD; BHD skin lesion fibrofolliculoma protein; Birt Hogg Dube syndrome protein; FLCL; Folliculin; FLCN_HUMAN.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Chicken,Dog,Cow,Horse,Sheep,
Applications:	ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800IF=1:100-500(Paraffin sections need antigen repair) not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	64kDa
Cellular localization:	The nucleuscytoplasmic
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human FLCN/BHD:65-165/579
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Storage:	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20°C. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.
PubMed:	PubMed
Product Detail:	This gene is located within the Smith-Magenis syndrome region on chromosome 17. Mutations in this gene are associated with Birt-Hogg-Dube syndrome, which is characterized by fibrofolliculomas, renal tumors, lung cysts, and pneumothorax. Alternative splicing of this gene results in three transcript variants encoding different isoforms.

Function:
May play a role in the pathogenesis of an uncommon form of kidney cancer through its association with an inherited disorder of the hair follicle (fibrofolliculomas). May be a tumor suppressor. May be involved in colorectal tumorigenesis. May be involved in energy and/or nutrient sensing through the AMPK and mTOR signaling pathways. May regulate phosphorylation of RPS6KB1.

Subunit:
Interacts (via C-terminus) with FNIP1 and FNIP2 (via C-terminus). This mediates indirect interaction with the PRKAA1, PRKAB1 and PRKAG1 subunits of 5'-AMP-activated protein kinase.

Subcellular Location:
Cytoplasm. Nucleus. Note=Mainly localized in the nucleus. Co-localizes with FNIP1 and FNIP2 in the cytoplasm.

Tissue Specificity:
Expressed in most tissues tested, including skin, lung, kidney, heart, testis and stomach.

Post-translational modifications:
Phosphorylated. Several different phosphorylated forms exist.

DISEASE:
Defects in FLCN are the cause of Birt-Hogg-Dube syndrome (BHD) [MIM:135150]. BHD is a rare autosomal dominant genodermatosis characterized by hair follicle hamartomas (fibrofolliculomas), kidney tumors, and spontaneous pneumothorax. Fibrofolliculomas are part of the triad of BHD skin lesions that also includes trichodiscomas and acrochordons. Onset of this dermatologic condition is invariably in adulthood. BHD is associated with a variety of histologic types of renal tumors, including chromophobe renal cell carcinoma (RCC), benign renal oncocytoma, clear-cell RCC and papillary type I RCC. Multiple lipomas, angiolipomas, and parathyroid adenomas are also seen in patients affected with this disease. The majority of mutations are predicted to prematurely terminate the protein.
Defects in FLCN are in some cases a cause of primary spontaneous pneumothorax (PSP) [MIM:173600]. PSP is a condition in which air is present in the pleural space in the absence of a precipitating event, such as trauma or lung disease. This results in secondary collapse of the lung, either partially or completely, and some degree of hypoxia. PSP is relatively common, with an incidence between 7.4-18/100'000 for men and 1.2-6/100'000 for women and a dose-dependent, increased risk among smokers. Most cases are sporadic, typically occurring in tall, thin men aged 10-30 years and generally while at rest. Familial PSP is rarer and usually is inherited as an autosomal dominant condition with reduced penetrance, although X-linked recessive and autosomal recessive inheritance have also been suggested.
Defects in FLCN may be a cause of renal cell carcinoma (RCC) [MIM:144700]. Renal cell carcinoma is a heterogeneous group of sporadic or hereditary carcinoma derived from cells of the proximal renal tubular epithelium. It is subclassified into clear cell

renal carcinoma (non-papillary carcinoma), papillary renal cell carcinoma, chromophobe renal cell carcinoma, collecting duct carcinoma with medullary carcinoma of the kidney, and unclassified renal cell carcinoma. Clear cell renal cell carcinoma is the most common subtype.

Similarity:
Belongs to the folliculin family.

SWISS:
Q8NFG4

Gene ID:
201163

Database links:

Entrez Gene: 201163Human

Omim: 607273Human

SwissProt: Q8NFG4Human

Unigene: 31652Human

Important Note:
This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.

BHD综合征(Birt-Hogg-Dube综合征)是指:患者患有多发性纤维毛囊瘤，并伴发软垂疣、胶原瘤、脂肪瘤和(或)口腔纤维瘤。是一种显性遗传综合征，有报告BHD患者可伴发髓样癌、结肠癌和肾细胞癌。
BHD综合征，主要为纤维毛囊瘤约2～4mm大小，皮肤色丘疹，可单个存在，但常见为多发性，散在分布于颜面、躯干和四肢。另外可伴发软垂疣、胶原瘤、脂肪瘤和(或)口腔纤维瘤。