

Cat. No:	ABN19790
Conjugate:	Unconjugated
Size:	100 μ L
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	The antiserum was produced against synthesized peptide derived from the N-terminal region of human VHL. AA range:1-50
Reactivity:	Human,Rat,Mouse
Applications:	IHC 1:50-1:200,ICC/IF 1:50-1:200,ELISA 1:10000-1:20000
Molecular Weight:	19-24kDa
Purification:	Affinity purification
Synonyms:	Von Hippel-Lindau disease tumor suppressor (Protein G7;pVHL)

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Background:

von Hippel-Lindau tumor suppressor(VHL) Homo sapiens Von Hippel-Lindau syndrome (VHL) is a dominantly inherited familial cancer syndrome predisposing to a variety of malignant and benign tumors. A germline mutation of this gene is the basis of familial inheritance of VHL syndrome. The protein encoded by this gene is a component of the protein complex that includes elongin B, elongin C, and cullin-2, and possesses ubiquitin ligase E3 activity. This protein is involved in the ubiquitination and degradation of hypoxia-inducible-factor (HIF), which is a transcription factor that plays a central role in the regulation of gene expression by oxygen. RNA polymerase II subunit POLR2G/RPB7 is also reported to be a target of this protein. Alternatively spliced transcript variants encoding distinct isoforms have been observed. [provided by RefSeq, Jul 2008],disease:Defects in VHL are a cause of pheochromocytoma [MIM:171300]. The pheochromocytomas are catecholamine-producing, chromaffin tumors that arise in the adrenal medulla in 90% of cases. In the remaining 10% of cases, they develop in extra-adrenal sympathetic ganglia and may be referred to as "paraganglioma." Pheochromocytoma usually presents with hypertension. Approximately 10% of pheochromocytoma is hereditary. The genetic basis for most cases of non-syndromic familial pheochromocytoma is unknown.,disease:Defects in VHL are a cause of renal cell carcinoma type 1 (RCC1) [MIM:144700]; also called hypernephroma or adenocarcinoma of kidney. Familial renal cell carcinoma syndromes form a group of diseases characterized by a predisposition to development of renal cell carcinomas (RCCs) with various histological subtypes.,disease:Defects in VHL are the cause of erythrocytosis familial type 2 (ECYT2) [MIM:263400]; also called VHL-dependent polycythemia or Chuvash type polycythemia. ECYT2 is an autosomal recessive disorder characterized by an increase in serum red blood cell mass, hypersensitivity of erythroid progenitors to erythropoietin, increased erythropoietin serum levels, and normal oxygen affinity. Patients with ECYT2 carry a high risk for peripheral thrombosis and cerebrovascular events.,disease:Defects in VHL are the cause of von Hippel-Lindau disease (VHLD) [MIM:193300]. VHLD is a dominantly inherited familial cancer syndrome characterized by the development of retinal angiomas, cerebellar and spinal hemangioblastoma, renal cell carcinoma (RCC), phaeochromocytoma and pancreatic tumors. VHL type 1 is without pheochromocytoma, type 2 is with pheochromocytoma. VHL type 2 is further subdivided into types 2A

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