Recombinant Human Von Hippel Lindau/VHL protein ab82240

Description

Product name: Recombinant Human Von Hippel Lindau/VHL protein
Purity: > 90 % SDS-PAGE. ab82240 is greater than 90% homogeneous based on SDS-PAGE analysis, purified using affinity and FPLC chromatography under non-denaturing conditions.
Expression system: Baculovirus
Protein length: Full length protein
Animal free: No
Nature: Recombinant
Species: Human

Specifications

Our Abpromise guarantee covers the use of ab82240 in the following tested applications.
The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.
Applications: SDS-PAGE, Western blot, Other
Form: Liquid

Preparation and Storage

Stability and Storage: Shipped on dry ice. Upon delivery aliquot and store at -80°C. Avoid freeze / thaw cycles.
  pH: 8.00
  Constituents: 0.75% Potassium chloride, 0.0154% DTT, 0.316% Tris HCl, 0.00584% EDTA, 20% Glycerol

General Info

Function: Involved in the ubiquitination and subsequent proteasomal degradation via the von Hippel-Lindau ubiquitination complex. Seems to act as target recruitment subunit in the E3 ubiquitin ligase
complex and recruits hydroxylated hypoxia-inducible factor (HIF) under normoxic conditions. Involved in transcriptional repression through interaction with HIF1A, HIF1AN and histone deacetylases.

**Tissue specificity**
Expressed in the adult and fetal brain and kidney.

**Pathway**
Protein modification; protein ubiquitination.

**Involvement in disease**
Defects in VHL are a cause of susceptibility to pheochromocytoma (PCC) [MIM:171300]. A catecholamine-producing tumor of chromaffin tissue of the adrenal medulla or sympathetic paraganglia. The cardinal symptom, reflecting the increased secretion of epinephrine and norepinephrine, is hypertension, which may be persistent or intermittent. 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 angiomatosis, 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 (pheochromocytoma, retinal angioma, and hemangioblastomas without renal cell carcinoma and pancreatic cyst) and 2B (pheochromocytoma, retinal angioma, and hemangioblastomas with renal cell carcinoma and pancreatic cyst). VHL type 2C refers to patients with isolated pheochromocytoma without hemangioblastoma or renal cell carcinoma. The estimated incidence is 3/100000 births per year and penetrance is 97% by age 60 years. 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. Defects in VHL are 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.

**Domain**
The Elongin BC complex binding domain is also known as BC-box with the consensus [APST]-L-x(3)-C-x(3)-[AILV].

**Cellular localization**

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Please note: All products are "FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC PROCEDURES"

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