abcam

Product datasheet

Recombinant Human Hamartin protein ab152772

1 Image

Description

Product name	Recombinant Human Hamartin protein
Expression system	Wheat germ
Accession	Q92574
Protein length	Protein fragment
Animal free	No
Nature	Recombinant
Species	Human
Sequence	LKKPGHVAEVYLVHLHASVYALFHRLYGMYPCNFVSFLRS HYSMKENLET FEEVVKPMMEHVRIHPELVTGSKDHELDPRRWKRLETHD VVIECAKISLD PTEASYEDG
Predicted molecular weight	38 kDa including tags
Amino acids	166 to 274

Specifications

Our **Abpromise guarantee** covers the use of **ab152772** in the following tested applications.

The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

Applications	Western blot SDS-PAGE ELISA
Form	Liquid
Additional notes	

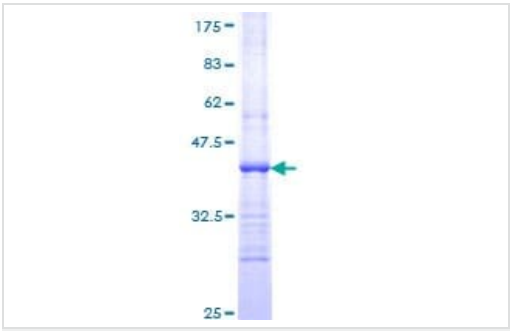
Preparation and Storage

Stability and Storage	Shipped on dry ice. Upon delivery aliquot and store at -80ºC. Avoid freeze / thaw cycles. pH: 8.00 Constituents: 0.31% Glutathione, 0.79% Tris HCl

General Info

Function	In complex with TSC2, inhibits the nutrient-mediated or growth factor-stimulated phosphorylation of S6K1 and EIF4EBP1 by negatively regulating mTORC1 signaling. Seems not to be required for TSC2 GAP activity towards RHEB. Implicated as a tumor suppressor. Involved in microtubule-mediated protein transport, but this seems to be due to unregulated mTOR signaling.
Tissue specificity	Highly expressed in skeletal muscle, followed by heart, brain, placenta, pancreas, lung, liver and kidney. Also expressed in embryonic kidney cells.
Involvement in disease	Defects in TSC1 are the cause of tuberous sclerosis type 1 (TSC1) [MIM:191100]. It is an autosomal dominant multi-system disorder that affects especially the brain, kidneys, heart, and skin. TS1C is characterized by hamartomas (benign overgrowths predominantly of a cell or tissue type that occurs normally in the organ) and hamartias (developmental abnormalities of tissue combination). Clinical symptoms can range from benign hypopigmented macules of the skin to profound mental retardation with intractable seizures to premature death from a variety of disease-associated causes. Defects in TSC1 may be a cause of focal cortical dysplasia of Taylor balloon cell type (FCDBC) [MIM:607341]. FCDBC is a subtype of cortical displasias linked to chronic intractable epilepsy. Cortical dysplasias display a broad spectrum of structural changes, which appear to result from changes in proliferation, migration, differentiation, and apoptosis of neuronal precursors and neurons during cortical development.
Domain	The C-terminal putative coiled-coil domain is necessary for interaction with TSC2.
Post-translational modifications	Phosphorylation at Ser-505 does not affect interaction with TSC2. Phosphorylated upon DNA damage, probably by ATM or ATR.
Cellular localization	Cytoplasm. Membrane. At steady state found in association with membranes.

Images



SDS-PAGE - Recombinant Human Hamartin protein (ab152772)

12.5% SDS-PAGE analysis of ab152772 stained with Coomassie Blue.

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