abcam

Product datasheet

Recombinant Human Hamartin protein ab152772

画像数1

製品の詳細

製品名 Recombinant Human Hamartin protein

発現系Wheat germアクセッション番号Q92574

タンパク質長 Protein fragment

Animal free No

由来 Recombinant

生物種 Human

配列 LKKPGHVAEVYLVHLHASVYALFHRLYGMYPCNFVSFLRSHY

SMKENLET

 ${\tt FEEVVKPMMEHVRIHPELVTGSKDHELDPRRWKRLETHDVVI}$

ECAKISLD PTEASYEDG

予測される分子量 38 kDa including tags

領域 166 to 274

特性

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.

アプリケーション Western blot

SDS-PAGE

ELISA

製品の状態 Liquid

備考

前処理および保存

保存方法および安定性 Shipped on dry ice. Upon delivery aliquot and store at -80°C. Avoid freeze / thaw cycles.

pH: 8.00

Constituents: 0.31% Glutathione, 0.79% Tris HCI

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関連情報

機能

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.

組織特異性

Highly expressed in skeletal muscle, followed by heart, brain, placenta, pancreas, lung, liver and kidney. Also expressed in embryonic kidney cells.

関連疾患

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.

ドメイン

The C-terminal putative coiled-coil domain is necessary for interaction with TSC2.

翻訳後修飾

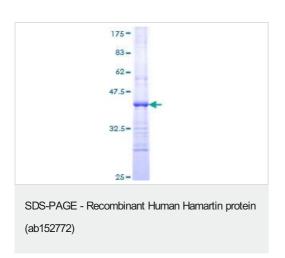
Phosphorylation at Ser-505 does not affect interaction with TSC2. Phosphorylated upon DNA

damage, probably by ATM or ATR.

細胞内局在

Cytoplasm. Membrane. At steady state found in association with membranes.

画像



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

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