Hamartin Rabbit pAb

CatalogNo: YT5760

Key Features

Host Species
- Rabbit

Reactivity
- Human,Rat,Mouse,

Applications
- WB,ELISA

MW
- 130kD (Observed)

Isotype
- IgG

Recommended Dilution Ratios

WB 1:500-2000
ELISA 1:10000-20000

Storage

Storage*	-15°C to -25°C/1 year(Do not lower than -25°C)
Formulation	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% sodium azide.

Basic Information

Clonality	Polyclonal

Immunogen Information

Immunogen	Synthesized peptide derived from Hamartin . at AA range: 360-440
Specificity	Hamartin Polyclonal Antibody detects endogenous levels of Hamartin

Target Information

Gene name	TSC1 KIAA0243 TSC

Protein Name Hamartin

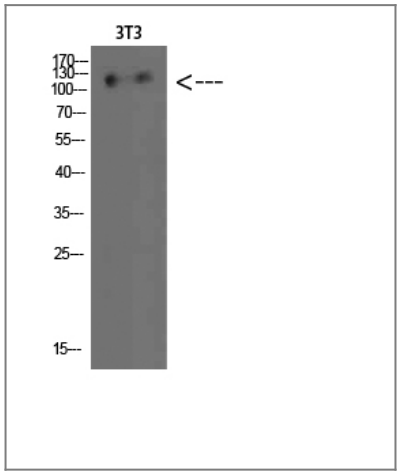
Organism	Gene ID	UniProt ID
Human	7248;	Q92574;
Mouse	64930;	Q9EP53;

Cellular Localization Cytoplasm . Membrane ; Peripheral membrane protein . At steady state found in association with membranes. .

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

Function Disease:Defects in TSC1 are the cause of tuberous sclerosis complex (TSC) [MIM:191100]. The molecular basis of TSC is a functional impairement of the hamartin-tuberin complex. TSC is an autosomal dominant multi-system disorder that affects especially the brain, kidneys, heart, and skin. TSC 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.,Disease: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.,Function:Implicated as a tumor suppressor. May have a function in vesicular transport. Interaction between TSC1 and TSC2 may facilitate vesicular docking.,PTM:Phosphorylated upon DNA damage, probably by ATM or ATR.,PTM:Phosphorylation at Ser-505 does not affect interaction with TSC2.,subcellular location:At steady state found in association with membranes.,subunit:Interacts with TSC2, leading to stabilize TSC2. In the absence of TSC2, TSC1 self-aggregates. Interacts with DOCK7.,tissue specificity:Highly expressed in skeletal muscle, followed by heart, brain, placenta, pancreas, lung, liver and kidney. Also expressed in embryonic kidney cells.,

Validation Data



Western Blot analysis of 3T3 cells using Hamartin Polyclonal Antibody diluted at 1:500. Secondary antibody(catalog#:RS0002) was diluted at 1:20000

Contact information

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Please scan the QR code to access additional product information:
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