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Melanophilin Rabbit pAb

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Catalog # AP57248

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	Q9BV36
Reactivity	Rat
Predicted	Human, Mouse
Host	Rabbit
Clonality	Polyclonal
Calculated MW	65949
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human Melanophilin
Epitope Specificity	451-550/600
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SUBCELLULAR LOCATION	Cytoplasm.
SIMILARITY	Contains 1 FYVE-type zinc finger. Contains 1 RabBD (Rab-binding) domain.
DISEASE	Defects in MLPH are a cause of Griscelli syndrome type 3 (GS3) [MIM:609227]. GS3 is a rare autosomal recessive disorder characterized by pigmentary dilution of the skin and hair, the presence of large clumps of pigment in hair shafts, and an accumulation of melanosomes in melanocytes, without other clinical manifestations.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	This gene encodes a member of the exophilin subfamily of Rab effector proteins. The protein forms a ternary complex with the small Ras-related GTPase Rab27A in its GTP-bound form and the motor protein myosin Va. A similar protein complex in mouse functions to tether pigment-producing organelles called melanosomes to the actin cytoskeleton in melanocytes, and is required for visible pigmentation in the hair and skin. A mutation in this gene results in Griscelli syndrome type 3, which is characterized by a silver-gray hair color and abnormal pigment distribution in the hair shaft. Several alternatively spliced transcript variants encoding different isoforms have been found for this gene. [provided by RefSeq, Jul 2013]

Additional Information

Gene ID	79083
Other Names	Melanophilin, Exophilin-3, Slp homolog lacking C2 domains a, SlaC2-a, Synaptotagmin-like protein 2a, MLPH, SLAC2A
Dilution	IHC-P=1:100-500,IHC-F=1:100-500,IF=1:100-500

Storage	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.

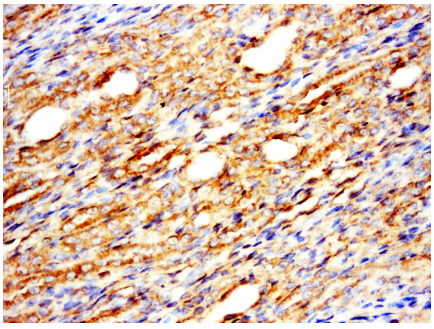
Protein Information

Name	MLPH
Synonyms	SLAC2A
Function	Rab effector protein involved in melanosome transport. Serves as link between melanosome-bound RAB27A and the motor protein MYO5A.
Cellular Location	Cytoplasm.

Background

This gene encodes a member of the exophilin subfamily of Rab effector proteins. The protein forms a ternary complex with the small Ras-related GTPase Rab27A in its GTP-bound form and the motor protein myosin Va. A similar protein complex in mouse functions to tether pigment-producing organelles called melanosomes to the actin cytoskeleton in melanocytes, and is required for visible pigmentation in the hair and skin. A mutation in this gene results in Griscelli syndrome type 3, which is characterized by a silver-gray hair color and abnormal pigment distribution in the hair shaft. Several alternatively spliced transcript variants encoding different isoforms have been found for this gene. [provided by RefSeq, Jul 2013]

Images



Paraformaldehyde-fixed, paraffin embedded (rat kidney); Antigen retrieval by boiling in sodium citrate buffer (pH6.0) for 15min; Block endogenous peroxidase by 3% hydrogen peroxide for 20 minutes; Blocking buffer (normal goat serum) at 37°C for 30min; Antibody incubation with (Melanophilin) Polyclonal Antibody, Unconjugated (AP57248) at 1:400 overnight at 4°C, followed by a conjugated secondary (sp-0023) for 20 minutes and DAB staining.