10320 Camino Santa Fe, Suite G
San Diego, CA 92121
Tel: 858.875.1900 Fax: 858.875.1999

MYO5A Rabbit pAb

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Catalog # AP57426

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	Q9Y4I1
Predicted	Rat, Pig, Dog, Human, Mouse, Rabbit, Chicken, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	215405
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human MYO5A
Epitope Specificity	1301-1400/1855
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
DISEASE	Defects in MYO5A are a cause of Griscelli syndrome type 1 (GS1) [MIM:214450]; also known as Griscelli syndrome with primary neurologic impairment. Griscelli syndrome is a rare autosomal recessive disorder that results in pigmentary dilution of the skin and hair, the presence of large clumps of pigment in hair shafts, silvery-gray hair and accumulation of melanosomes in melanocytes. GS1 patients show developmental delay, hypotonia and mental retardation, without apparent immune abnormalities. Defects in MYO5A are a cause of Griscelli syndrome type 3 (GS3) [MIM:609227]. GS3 is characterized by pigmentary dilution of the skin and hair, the presence of large clumps of pigment in hair shafts, silvery-gray hair and accumulation of melanosomes in melanocytes, without other clinical manifestations. Defects in MYO5A are a cause of Elejalde syndrome (ELEJAS) [MIM:256710]; also known as neuroectodermal melanolysosomal disease. Elejalde syndrome is an autosomal recessive condition characterized by skin hypopigmentation, the presence of large clumps of pigment in hair shafts, silvery-gray hair, accumulation of melanosomes in melanocytes and primary neurological abnormalities. Elejalde syndrome may be the same entity as Griscelli syndrome type 1.
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 is one of three myosin V heavy-chain genes, belonging to the myosin gene superfamily. Myosin V is a class of actin-based motor proteins involved in cytoplasmic vesicle transport and anchorage, spindle-pole alignment and mRNA translocation. The protein encoded by this gene is abundant in melanocytes and nerve cells. Mutations in this gene cause Griscelli syndrome type-1 (GS1), Griscelli syndrome type-3 (GS3) and neuroectodermal melanolysosomal disease, or Elejalde disease. Multiple alternatively spliced transcript variants encoding different isoforms have been reported, but the full-length nature of some variants has not been determined. [provided by RefSeq, Dec 2008]

Additional Information

Gene ID	4644
Other Names	Unconventional myosin-Va, Dilute myosin heavy chain, non-muscle, Myosin heavy chain 12, Myosin-12, Myoxin, MYO5A, MYH12
Dilution	IHC-P=1:100-500,IHC-F=1:100-500,ICC/IF=1:100-500,IF=1:100-500,ELISA=1:5000-10000
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	MYO5A
Synonyms	MYH12
Function	Processive actin-based motor that can move in large steps approximating the 36-nm pseudo-repeat of the actin filament. Can hydrolyze ATP in the presence of actin, which is essential for its function as a motor protein (PubMed:10448864). Involved in melanosome transport. Also mediates the transport of vesicles to the plasma membrane (By similarity). May also be required for some polarization process involved in dendrite formation (By similarity).
Tissue Location	Detected in melanocytes.

Background

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