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Rabbit Anti-MYO5A/AP Conjugated antibody

SL19173R-AP

Product Name	Anti-MYO5A/AP
Chinese Name	碱性磷酸酶（AP）标记的肌球蛋白 5A 抗体
Alias	Dilute myosin heavy chain; GS1; MYH12; MYO5; Myo5a; MYO5A_HUMAN; Myosin heavy chain 12; Myosin heavy polypeptide kinase; Myosin V; Myosin VA (heavy polypeptide 12 myoxin); Myosin-12; Myosin-Va; Myoxin; MYR12; non-muscle.
Research Area	Cell biology immunology Neurobiology Signal transduction
Immunogen Species	Rabbit
Clonality	Polyclonal
React Species	(predicted:Human,Mouse,Rat,Chicken,Dog,Pig,Cow,Horse,Rabbit,Sheep)
Applications	IHC-P=1:100-500,IHC-F=1:100-500,ELISA=1:500-5000 not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight	215kDa
Form	Lyophilized or Liquid
Concentration	1mg/ml
immunogen	KLH conjugated synthetic peptide derived from human MYO5A
Lsotype	IgG
Purification	affinity purified by Protein A
Storage Buffer	1M TBS(pH7.4) with 1% BSA, 3% Proclin300 and 50% Glycerol.
Storage	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20°C. When reconstituted in sterile pH 7.4 1M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.
Product Detail	**background:** 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]

Function:
Processive actin-based motor that can move in large steps approximating the 36-nm pseudo-repeat of the actin filament. Involved in melanosome transport. May also be required for some polarization process involved in dendrite formation.

Tissue Specificity:
Detected in melanocytes.

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.

Similarity:
Contains 1 dilute domain.
Contains 6 IQ domains.
Contains 1 myosin head-like domain.

Database links:

Entrez Gene: 4644 Human

Entrez Gene: 17918 Mouse

Entrez Gene: 594849 Pig

Entrez Gene: 25017 Rat

Omim: 160777 Human

SwissProt: Q02440 Chicken

SwissProt: Q9Y4I1 Human

SwissProt: Q99104 Mouse

SwissProt: Q9QYF3 Rat

Unigene: 21213 Human

Unigene: 596221 Human

Unigene: 3645 Mouse

Unigene: 44865 Rat

Important Note:
This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.