

Product name:	Myosin VA Rabbit Polyclonal Antibody
Cat number:	ABN14346
Conjugate:	Unconjugated
Size:	100µL
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	The antiserum was produced against synthesized peptide derived from human MYO5A. AA range:1784-1833
Reactivity:	Human,Mouse,Rat
Applications:	WB 1:500-1:2000,IHC 1:50-1:300
Molecular Weight:	220kDa
Purification:	Affinity purification
Form:	Liquid
Buffer:	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% New type preservative N.
Storage:	Store at 4°C short term. Aliquot and store at -20°C for 12 months. Avoid freeze/thaw cycles.

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],disease:Defects in MYO5A are a cause of Elejalde syndrome [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 I.,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.,disease: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.,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.,online information:MYO5A mutation db,similarity:Contains 1 dilute domain.,similarity:Contains 1 myosin head-like domain.,similarity:Contains 6 IQ domains.,subunit:May be a homodimer, which associates with multiple calmodulin or myosin light chains. Binds MLPH and MYRIP.,tissue specificity:Detected in melanocytes.,