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Chorein Rabbit pAb

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Catalog # AP55344

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	Q96RL7
Predicted	Rat, Human, Mouse, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	360276
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human Chorein
Epitope Specificity	3001-3174/3174
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
DISEASE	Choreoacanthocytosis (CHAC) [MIM:200150]: An autosomal recessive neurodegenerative disorder characterized by the gradual onset of hyperkinetic movements and abnormal erythrocyte morphology. Basal ganglia atrophy in the brain is a pathological feature of the disease. Other clinical symptoms include psychiatric features, epilepsy, peripheral neuropathy, myopathy and oral self-mutilation. Note=The disease is caused by mutations affecting the gene represented in this entry.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	Chorein may control steps in the cycling of proteins through the trans-Golgi network to endosomes, lysosomes and the plasma membrane. Mutations in this gene cause the autosomal recessive disorder chorea-acanthocytosis. Alternative splicing of this gene results in multiple transcript variants.

Additional Information

Gene ID	23230
Other Names	Intermembrane lipid transfer protein VPS13A, Chorea-acanthocytosis protein, Chorein, Vacuolar protein sorting-associated protein 13A, VPS13A, CHAC, KIAA0986
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	VPS13A
Synonyms	CHAC, KIAA0986
Function	Mediates the transfer of lipids between membranes at organelle contact sites (By similarity). Binds phospholipids (PubMed:34830155). Required for the formation or stabilization of ER- mitochondria contact sites which enable transfer of lipids between the ER and mitochondria (PubMed:30741634). Negatively regulates lipid droplet size and motility (PubMed:30741634). Required for efficient lysosomal protein degradation (PubMed:30709847).
Cellular Location	Mitochondrion outer membrane; Peripheral membrane protein. Endoplasmic reticulum membrane; Peripheral membrane protein. Endosome membrane; Peripheral membrane protein. Lysosome membrane; Peripheral membrane protein. Lipid droplet. Golgi apparatus {ECO:0000250|UniProtKB:Q5H8C4}. Cytoplasmic vesicle, secretory vesicle, neuronal dense core vesicle {ECO:0000250|UniProtKB:Q5H8C4}. Note=Localizes at mitochondria-endosomes and mitochondria-endoplasmic reticulum contact sites (PubMed:30093493, PubMed:30709847, PubMed:30741634)
Tissue Location	Expressed in red blood cells (at protein level) (PubMed:31086825). Widely expressed, with high expression in brain, heart, skeletal muscle and kidney (PubMed:15498460)

Background

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