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FAM126A Rabbit pAb

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Catalog # AP54542

Product Information

Application	WB
Primary Accession	Q9BYI3
Reactivity	Human, Mouse
Predicted	Rat, Dog, Pig, Horse, Rabbit
Host	Rabbit
Clonality	Polyclonal
Calculated MW	57625
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human FAM126A
Epitope Specificity	1-100/521
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. Membrane. According to PubMed:10910037, it is mainly cytoplasmic while according to PubMed:16951682, it is a membrane protein.
SIMILARITY	Belongs to the FAM126 family.
DISEASE	Defects in FAM126A are the cause of leukodystrophy hypomyelinating type 5 (HLD5) [MIM:610532]. This disorder is characterized by congenital cataract, progressive neurologic impairment, and diffuse myelin deficiency. Affected individuals experience progressive pyramidal and cerebellar dysfunction, muscle weakness and wasting prevailingly in the lower limbs. Mental deficiency ranges from mild to moderate.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	Hyccin is a 521 amino acid cytoplasmic protein that is widely expressed with highest levels found in heart, brain, placenta, spleen and testis. Belonging to the FAM126 family, hyccin may play a role in the ∫-catenin/Lef signaling pathway. Hyccin is likely involved in the process of myelination of the central and peripheral nervous system. Defects in the gene encoding hyccin are the cause of leukodystrophy hypomyelinating type 5 (HLD5), which is characterized by congenital cataract, progressive neurologic impairment and diffuse myelin deficiency. Individuals affected by HLD5 experience progressive pyramidal and cerebellar dysfunction along with muscle weakness in the lower limbs. Hyccin exists as two alternatively spliced isoforms and is encoded by a gene located on human chromosome 7.

Additional Information

Gene ID	84668
Other Names	Hyccin, Down-regulated by CTNNB1 protein A, HYCC1 (HGNC:24587)

Target/Specificity	Widely expressed. Highest levels in heart, brain, placenta, spleen and testis.
Dilution	WB=1:500-2000
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.

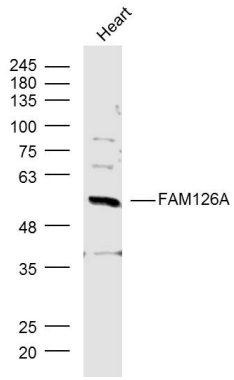
Protein Information

Name	HYCC1 (HGNC:24587)
Function	Component of a complex required to localize phosphatidylinositol 4-kinase (PI4K) to the plasma membrane (PubMed:26571211). The complex acts as a regulator of phosphatidylinositol 4-phosphate (PtdIns(4)P) synthesis (PubMed:26571211). HYCC1 plays a key role in oligodendrocytes formation, a cell type with expanded plasma membrane that requires generation of PtdIns(4)P (PubMed:26571211). Its role in oligodendrocytes formation probably explains its importance in myelination of the central and peripheral nervous system (PubMed:16951682, PubMed:26571211). May also have a role in the beta- catenin/Lef signaling pathway (Probable).
Cellular Location	Cytoplasm, cytosol. Cell membrane Note=Localizes to the cytosol and is recruited to the plasma membrane following interaction with other components of the phosphatidylinositol 4-kinase (PI4K) complex.
Tissue Location	Widely expressed. Highest levels in heart, brain, placenta, spleen and testis.

Background

Hyccin is a 521 amino acid cytoplasmic protein that is widely expressed with highest levels found in heart, brain, placenta, spleen and testis. Belonging to the FAM126 family, hyccin may play a role in the ∫-catenin/Lef signaling pathway. Hyccin is likely involved in the process of myelination of the central and peripheral nervous system. Defects in the gene encoding hyccin are the cause of leukodystrophy hypomyelinating type 5 (HLD5), which is characterized by congenital cataract, progressive neurologic impairment and diffuse myelin deficiency. Individuals affected by HLD5 experience progressive pyramidal and cerebellar dysfunction along with muscle weakness in the lower limbs. Hyccin exists as two alternatively spliced isoforms and is encoded by a gene located on human chromosome 7.

Images



Sample:
Heart (Mouse) Lysate at 40 ug
Primary: Anti-FAM126A (AP54542) at 1/300 dilution
Secondary: IRDye800CW Goat Anti-Rabbit IgG at 1/20000 dilution
Predicted band size: 58 kD
Observed band size: 58 kD