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LMBRD1 Rabbit pAb

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Catalog # AP57032

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	Q9NUN5
Reactivity	Mouse
Predicted	Rat, Pig, Dog, Human, Rabbit, Chicken, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	61389
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human LMBRD1
Epitope Specificity	21-120/540
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	Lysosome membrane.
DISEASE	Defects in LMBRD1 are the cause of methylmalonic aciduria and homocystinuria type cblF (MMAFHC) [MIM:277380]; also known as homocystinuria-megaloblastic anemia complementation type F. MMAFHC is a disorder of cobalamin metabolism characterized by decreased levels of the coenzymes adenosylcobalamin (AdoCbl) and methylcobalamin (MeCbl). It is due to accumulation of free cobalamin in lysosomes, thus hindering its conversion to cofactors. Clinical features include developmental delay, stomatitis, glossitis, seizures and methylmalonic aciduria responsive to vitamin B12.
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 encodes a lysosomal membrane protein that may be involved in the transport and metabolism of cobalamin. This protein also interacts with the large form of the hepatitis delta antigen and may be required for the nucleocytoplasmic shuttling of the hepatitis delta virus. Mutations in this gene are associated with the vitamin B12 metabolism disorder termed, homocystinuria-megaloblastic anemia complementation type F.[provided by RefSeq, Oct 2009]

Additional Information

Gene ID	55788
Other Names	Lysosomal cobalamin transport escort protein LMBD1, LMBD1, HDAg-L-interacting protein NESI, LMBR1 domain-containing protein 1, Nuclear export signal-interacting protein, LMBRD1 (HGNC:23038), C6orf209, NESI

Dilution	IHC-P=1:100-500,IHC-F=1:100-500,IF=1:100-500
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	LMBRD1 (HGNC:23038)
Synonyms	C6orf209, NESI
Function	Lysosomal membrane chaperone required to export cobalamin (vitamin B12) from the lysosome to the cytosol, allowing its conversion to cofactors (PubMed:19136951). Targets ABCD4 transporter from the endoplasmic reticulum to the lysosome (PubMed:27456980). Then forms a complex with lysosomal ABCD4 and cytoplasmic MMACHC to transport cobalamin across the lysosomal membrane (PubMed:25535791). Acts as an adapter protein which plays an important role in mediating and regulating the internalization of the insulin receptor (INSR) (By similarity). Involved in clathrin-mediated endocytosis of INSR via its interaction with adapter protein complex 2 (By similarity). Essential for the initiation of gastrulation and early formation of mesoderm structures during embryogenesis (By similarity).
Cellular Location	Endoplasmic reticulum membrane. Lysosome membrane; Multi-pass membrane protein. Cell membrane {ECO:0000250|UniProtKB:Q8K0B2}; Multi-pass membrane protein. Cytoplasmic vesicle, clathrin-coated vesicle {ECO:0000250|UniProtKB:Q8K0B2}
Tissue Location	Isoform 3 is expressed in liver.

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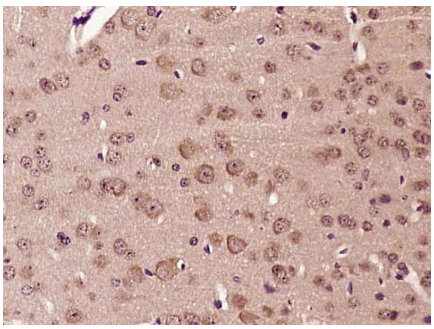
Images



Paraformaldehyde-fixed, paraffin embedded (Mouse brain); Antigen retrieval by boiling in sodium citrate buffer (pH6.0) for 15min; Block endogenous peroxidase by 3% hydrogen peroxide for 20 minutes; Blocking buffer (normal goat serum) at 37°C for 30min; Antibody incubation with (LMBRD1) Polyclonal Antibody, Unconjugated (AP57032) at 1:400 overnight at 4°C, followed by operating according to SP Kit(Rabbit) (sp-0023) instructionsand DAB staining.