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MLC1 Rabbit pAb

Catalog # AP98164

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	Q15049
Other Accession	Q15049
Reactivity	Rat, Mouse
Predicted	Pig, Human, Rabbit, Dog, Horse
Host	Rabbit
Clonality	Polyclonal
Calculated MW	41141
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human MLC1
Epitope Specificity	321-377/377
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SUBUNIT	Interacts with ATP1B1. Part of a complex containing ATP1B1, TRPV4, AQP4 and HEPACAM.
DISEASE	Defects in MLC1 are a cause of leukoencephalopathy megalencephalic with subcortical cysts (MLC) [MIM:604004]. MLC is a syndrome of cerebral leukoencephalopathy and megalencephaly characterized by ataxia, spasticity, seizures, delay in motor development and mild mental retardation. The brain appears swollen on magnetic resonance imaging, with diffuse white-matter abnormalities and the invariable presence of subcortical cysts in frontal and temporal lobes.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	MLC1 is a 377 amino acid multi-pass membrane protein that may serve as a non-selective neuronal cation channel in brain. Mutant MLC1 proteins that show impaired folding have been corrected in vitro with the addition of a Ca(2+)-ATPase inhibitor, curcumin. Mutations in the gene encoding MLC1 is the cause of megalencephalic leukoencephalopathy with subcortical cysts, also known as van der Knaap disease, a rare syndrome characterized early in life by progressive brain destruction causing mental retardation and incoordination. Single nucleotide polymorphisms within the MLC1 gene may be associated with periodic catatonia, but there seems to be conflicting evidence on whether or not the gene is implicated in general schizophrenia.

Additional Information

Gene ID	23209
Other Names	LVM; MLC; VL; Kiaa0027-hp; WKL1; MLC1_HUMAN; MLC1; Megalencephalic leukoencephalopathy with subcortical cysts protein 1; KIAA0027; MLC1_MOUSE; modulator of VRAC current 1; megalencephalic

	leukoencephalopathy with subcortical cysts 1
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	MLC1 (HGNC:17082)
Function	Transmembrane protein mainly expressed in brain astrocytes that may play a role in transport across the blood-brain and brain- cerebrospinal fluid barriers (PubMed:22328087). Regulates the response of astrocytes to hypo-osmosis by promoting calcium influx (PubMed:22328087). May function as regulatory protein of membrane protein complexes such as ion channels (Probable).
Cellular Location	Membrane; Multi-pass membrane protein. Cell membrane; Multi-pass membrane protein. Cytoplasm, perinuclear region. Endoplasmic reticulum
Tissue Location	Expressed in the brain, with highest levels found in the amygdala, nucleus caudatus, thalamus and hippocampus

Please note: All products are 'FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC OR THERAPEUTIC PROCEDURES'.