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PRRT2 Rabbit pAb

Catalog # AP96710

Product Information

Application	WB
Primary Accession	Q7Z6L0
Other Accession	Q7Z6L0
Reactivity	Mouse
Predicted	Pig, Human, Dog, Horse
Host	Rabbit
Clonality	Polyclonal
Calculated MW	34945
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human PRRT2
Epitope Specificity	201-300/340
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SIMILARITY	Belongs to the CD225/Dispanin family.
DISEASE	Episodic kinesigenic dyskinesia 1 (EKD1) [MIM:128200]: An autosomal dominant neurologic condition characterized by recurrent and brief attacks of abnormal involuntary movements, triggered by sudden voluntary movement. These attacks usually have onset during childhood or early adulthood and can involve dystonic postures, chorea, or athetosis. Note=The disease is caused by mutations affecting the gene represented in this entry. Disease-causing mutations that produce truncation of the C-terminus of the protein alter subcellular location, from plasma membrane to cytosplasm (PubMed:22101681). Convulsions, familial infantile, with paroxysmal choreoathetosis (ICCA) [MIM:602066]: A syndrome characterized by clinical features of benign familial infantile seizures and episodic kinesigenic dyskinesia. Benign familial infantile seizures is a disorder characterized by afebrile seizures occurring during the first year of life, without neurologic sequelae. Paroxysmal choreoathetosis is a disorder of involuntary movements characterized by attacks that occur spontaneously or are induced by a variety of stimuli. Note=The disease is caused by mutations affecting the gene represented in this entry. Seizures, benign familial infantile 2 (BFIS2) [MIM:605751]: An autosomal dominant disorder in which afebrile seizures occur in clusters during the first year of life, without neurologic sequelae. 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	This gene encodes a transmembrane protein containing a proline-rich domain in its N-terminal half. Studies in mice suggest that it is predominantly expressed in brain and spinal cord in embryonic and postnatal stages. Mutations in this gene are associated with episodic kinesigenic dyskinesia-1. Alternatively spliced transcript variants encoding different isoforms have been found for this gene. [provided by RefSeq, Jan 2012]

Additional Information

Gene ID	112476
Other Names	BFIC2; BFIS2; DSPB3; DYT10; EKD1; FICCA; ICCA; IFITMD1; PKC; 1500031I19Rik; RGD1564195; PRRT2_HUMAN; PRRT2; Dispanin subfamily B member 3 (DSPB3); PRRT2_MOUSE; PRRT2_RAT; proline rich transmembrane protein 2; infantile convulsions and paroxysmal choreoathetosis; dystonia 10; proline-rich transmembrane protein 2; interferon induced transmembrane protein domain containing 1; dispanin subfamily B member 3; Episodic kinesigenic dyskinesia 1; Paroxysmal kinesigenic dyskinesia
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	PRRT2
Function	As a component of the outer core of AMPAR complex, may be involved in synaptic transmission in the central nervous system. In hippocampal neurons, in presynaptic terminals, plays an important role in the final steps of neurotransmitter release, possibly by regulating Ca(2+)-sensing. In the cerebellum, may inhibit SNARE complex formation and down-regulate short-term facilitation.
Cellular Location	Cell membrane; Single-pass membrane protein {ECO:0000250|UniProtKB:E9PUL5}. Presynaptic cell membrane {ECO:0000250|UniProtKB:E9PUL5}; Single-pass membrane protein {ECO:0000250|UniProtKB:E9PUL5}. Synapse {ECO:0000250|UniProtKB:E9PUL5} Cell projection, axon. Cytoplasmic vesicle, secretory vesicle, synaptic vesicle membrane {ECO:0000250|UniProtKB:D3ZFB6}. Postsynaptic density membrane {ECO:0000250|UniProtKB:D3ZFB6}. Cell projection, dendritic spine {ECO:0000250|UniProtKB:D3ZFB6}

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