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LRRC8A Rabbit pAb

Catalog # AP97798

Product Information

Application	WB, IHC-P, IHC-F, IF, E
Primary Accession	Q8IWT6
Other Accession	Q8IWT6
Predicted	Rat, Pig, Human, Mouse, Rabbit, Dog, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	94199
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human LRRC8A
Epitope Specificity	84-180/810
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SIMILARITY	Contains 17 LRR (leucine-rich) repeats.
DISEASE	Defects in LRRC8A are the cause of agammaglobulinemia type 5 (AGM5) [MIM:613506]. It is a primary immunodeficiency characterized by profoundly low or absent serum antibodies and low or absent circulating B-cells due to an early block of B-cell development. Affected individuals develop severe infections in the first years of life. Note=A chromosomal aberration involving LRRC8 has been found in a patient with congenital agammaglobulinemia. Translocation t(9;20)(q33.2;q12). The translocation truncates the LRRC8 gene, resulting in deletion of the eighth, ninth, and half of the seventh LRR domains.
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 protein belonging to the leucine-rich repeat family of proteins, which are involved in diverse biological processes, including cell adhesion, cellular trafficking, and hormone-receptor interactions. This family member is a putative four-pass transmembrane protein that plays a role in B cell development. Defects in this gene cause autosomal dominant non-Bruton type agammaglobulinemia, an immunodeficiency disease resulting from defects in B cell maturation. Multiple alternatively spliced transcript variants, which encode the same protein, have been identified for this gene. [provided by RefSeq, Jul 2008]

Additional Information

Gene ID	56262
Other Names	LRC8A_HUMAN; LRRC8A; Leucine-rich repeat-containing protein 8A (HsLRRC8A); Swelling protein 1; KIAA1437; LRRC8; SWELL1; LRC8A_MOUSE; Leucine-rich repeat-containing protein 8A; Protein ebouriffe (ebo); LRC8A_RAT; Swelling protein 1 SWELL1; leucine rich repeat containing 8 VRAC subunit A; leucine rich repeat containing 8; leucine rich repeat containing 8

	family member A
Dilution	WB=1:500-2000,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	LRRC8A {ECO:0000303|PubMed:22532330, ECO:0000312|HGNC:HGNC:19027}
Function	Essential component of the volume-regulated anion channel (VRAC, also named VSOAC channel), an anion channel required to maintain a constant cell volume in response to extracellular or intracellular osmotic changes (PubMed:24725410, PubMed:24790029, PubMed:26530471, PubMed:26824658, PubMed:28193731, PubMed:29769723). The VRAC channel conducts iodide better than chloride and can also conduct organic osmolytes like taurine (PubMed:24725410, PubMed:24790029, PubMed:26530471, PubMed:26824658, PubMed:28193731, PubMed:30095067). Mediates efflux of amino acids, such as aspartate and glutamate, in response to osmotic stress (PubMed:28193731). LRRC8A and LRRC8D are required for the uptake of the drug cisplatin (PubMed:26530471). In complex with LRRC8C or LRRC8E, acts as a transporter of immunoreactive cyclic dinucleotide GMP-AMP (2'-3'-cGAMP), an immune messenger produced in response to DNA virus in the cytosol: mediates both import and export of 2'-3'-cGAMP, thereby promoting transfer of 2'-3'-cGAMP to bystander cells (PubMed:33171122). In contrast, complexes containing LRRC8D inhibit transport of 2'-3'-cGAMP (PubMed:33171122). Required for in vivo channel activity, together with at least one other family member (LRRC8B, LRRC8C, LRRC8D or LRRC8E); channel characteristics depend on the precise subunit composition (PubMed:24790029, PubMed:26824658, PubMed:28193731). Can form functional channels by itself (in vitro) (PubMed:26824658). Involved in B-cell development: required for the pro-B cell to pre-B cell transition (PubMed:14660746). Also required for T-cell development (By similarity). Required for myoblast differentiation: VRAC activity promotes membrane hyperpolarization and regulates insulin-stimulated glucose metabolism and oxygen consumption (By similarity). Also acts as a regulator of glucose-sensing in pancreatic beta cells: VRAC currents, generated in response to hypotonicity- or glucose-induced beta cell swelling, depolarize cells, thereby causing electrical excitation, leading to increase glucose sensitivity and insulin secretion (PubMed:29371604). Also plays a role in lysosome homeostasis by forming functional lysosomal VRAC channels in response to low cytoplasmic ionic strength condition: lysosomal VRAC channels are necessary for the formation of large lysosome-derived vacuoles, which store and then expel excess water to maintain cytosolic water homeostasis (PubMed:31270356, PubMed:33139539). Acts as a key factor in NLRP3 inflammasome activation by modulating itaconate efflux and mitochondria function (PubMed:39909992).
Cellular Location	Cell membrane; Multi-pass membrane protein. Lysosome membrane; Multi-pass membrane protein. Note=Mainly localizes to the cell membrane, with some intracellular localization to lysosomes
Tissue Location	Expressed in brain, kidney, ovary, lung, liver, heart, and fetal brain and liver. Found at high levels in bone marrow; lower levels are detected in peripheral

blood cells. Expressed on T- cells as well as on B-lineage cells.

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