

C9orf72 Antibody (C-term) Blocking peptide Synthetic peptide Catalog # BP12928b

Specification

C9orf72 Antibody (C-term) Blocking peptide - Product Information

Primary Accession Other Accession <u>Q96LT7</u> <u>NP 659442.2, NP 060795.1</u>

C9orf72 Antibody (C-term) Blocking peptide - Additional Information

Gene ID 203228

Other Names Protein C9orf72, C9orf72

Format

Peptides are lyophilized in a solid powder format. Peptides can be reconstituted in solution using the appropriate buffer as needed.

Storage

Maintain refrigerated at 2-8°C for up to 6 months. For long term storage store at -20°C.

Precautions This product is for research use only. Not for use in diagnostic or therapeutic procedures.

C9orf72 Antibody (C-term) Blocking peptide - Protein Information

Name C9orf72 (<u>HGNC:28337</u>)

Function

Acts as a guanine-nucleotide releasing factor (GEF) for Rab GTPases by promoting the conversion of inactive RAB-GDP to the active form RAB-GTP (PubMed:27103069, PubMed:27103069, PubMed:37821429). Acts as a GEF for RAB39A which enables HOPS-mediated autophagosome-lysosome membrane tethering and fusion in mammalian autophagy (PubMed:37821429). Component

of the C9orf72-SMCR8 complex where both subunits display GEF activity and that regulates autophagy (PubMed:<a href="http://www.uniprot.org/citations/27103069"

target="_blank">27103069, PubMed:27193190, PubMed:27617292, PubMed:28195531). As part of the C9orf72-SMCR8-WDR41 (CSW) complex, functions as GEF for RAB8A and RAB39B, thereby promoting autophagosome maturation



(PubMed:27103069). As part of the C9orf72-SMCR8 complex, also functions as GTPase activating protein (GAP) for RAB8A and RAB11A in vitro (PubMed:32303654). The C9orf72-SMCR8 complex also acts as a regulator of autophagy initiation by interacting with the ULK1/ATG1 kinase complex and modulating its protein kinase activity (PubMed:27617292). Promotes initiation of autophagy by regulating the RAB1A-dependent trafficking of the ULK1/ATG1 kinase complex to the phagophore which leads to autophagosome formation (PubMed:27334615). Acts as a regulator of mTORC1 signaling by promoting phosphorylation of mTORC1 substrates (PubMed:27559131). Plays a role

in endosomal trafficking (PubMed:24549040). May be involved in regulating the maturation of phagosomes to lysosomes (By similarity). Promotes the lysosomal localization and lysosome-mediated degradation of CARM1 which leads to inhibition of starvation-induced lipid metabolism (By similarity). Regulates actin dynamics in motor neurons by inhibiting the GTP-binding activity of ARF6, leading to ARF6 inactivation (PubMed: 27723745). This reduces the activity of the LIMK1 and LIMK2 kinases which are responsible for phosphorylation and inactivation of cofilin, leading to CFL1/cofilin activation (PubMed:27723745). Positively regulates axon extension and axon growth cone size in spinal motor neurons (PubMed:27723745). Required for SMCR8 protein expression and localization at pre- and post-synaptic compartments in the forebrain, also regulates protein abundance of RAB3A and GRIA1/GLUR1 in post-synaptic compartments in the forebrain and hippocampus (By similarity). Plays a role within the hematopoietic system in restricting inflammation and the development of autoimmunity (By similarity).

Cellular Location

Cytoplasm. Nucleus. Cytoplasm, P-body. Cytoplasm, Stress granule. Endosome Lysosome Cytoplasmic vesicle, autophagosome Autolysosome. Secreted. Cell projection, axon. Cell projection, growth cone. Perikaryon {ECO:0000250|UniProtKB:Q6DFW0}. Note=Detected in the cytoplasm of neurons from brain tissue (PubMed:21944778). Detected in the nucleus in fibroblasts (PubMed:21944779). During corticogenesis, transitions from being predominantly cytoplasmic to a more even nucleocytoplasmic distribution (By similarity). Majorly localized in cytosol under basal conditions (PubMed:37821429). Majorly gathered on autolysosomes structures under autophagy-induced conditions (PubMed:37821429) {ECO:0000250|UniProtKB:Q6DFW0, ECO:0000269|PubMed:21944778, ECO:0000269|PubMed:21944779,

ECO:0000269 PubMed:27037575, ECO:0000269 PubMed:37821429 [Isoform 2]: Nucleus membrane; Peripheral membrane protein. Nucleus. Note=Detected at the nuclear membrane of cerebellar Purkinje cells and spinal motor neurons. Also shows diffuse nuclear expression in spinal motor neurons

Tissue Location

Both isoforms are widely expressed, including kidney, lung, liver, heart, testis and several brain regions, such as cerebellum. Also expressed in the frontal cortex and in lymphoblasts (at protein level).

C9orf72 Antibody (C-term) Blocking peptide - Protocols

Provided below are standard protocols that you may find useful for product applications.

<u>Blocking Peptides</u>

C9orf72 Antibody (C-term) Blocking peptide - Images



C9orf72 Antibody (C-term) Blocking peptide - References

Suarez-Gestal, M., et al. Arthritis Res. Ther. 12 (2), R72 (2010) :van Es, M.A., et al. Nat. Genet. 41(10):1083-1087(2009)Humphray, S.J., et al. Nature 429(6990):369-374(2004)