

C9orf72 Antibody (C-term)

Affinity Purified Rabbit Polyclonal Antibody (Pab) Catalog # AP12928b

Specification

C9orf72 Antibody (C-term) - Product Information

Application WB,E
Primary Accession Q96LT7

Other Accession <u>Q66HC3</u>, <u>NP_060795.1</u>, <u>NP_659442.2</u>, <u>Q6DFW0</u>

Reactivity
Predicted
Mouse, Rat
Host
Clonality
Polyclonal
Isotype
Calculated MW
Antigen Region
Human
Mouse, Rat
Rabbit
Rabbit
Polyclonal
Rabbit IgG
396-424

C9orf72 Antibody (C-term) - Additional Information

Gene ID 203228

Other Names

Protein C9orf72, C9orf72

Target/Specificity

This C9orf72 antibody is generated from rabbits immunized with a KLH conjugated synthetic peptide between 396-424 amino acids from the C-terminal region of human C9orf72.

Dilution

WB~~1:1000

E~~Use at an assay dependent concentration.

Format

Purified polyclonal antibody supplied in PBS with 0.09% (W/V) sodium azide. This antibody is purified through a protein A column, followed by peptide affinity purification.

Storage

Maintain refrigerated at 2-8°C for up to 2 weeks. For long term storage store at -20°C in small aliquots to prevent freeze-thaw cycles.

Precautions

C9orf72 Antibody (C-term) is for research use only and not for use in diagnostic or therapeutic procedures.

C9orf72 Antibody (C-term) - Protein Information

Name C9orf72 (HGNC:28337)



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: <u>27193190</u>, PubMed: <u>27617292</u>, PubMed: <u>28195531</u>, PubMed: <u>37821429</u>). 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: 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) - Protocols

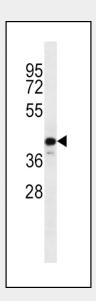
Provided below are standard protocols that you may find useful for product applications.

- Western Blot
- Blocking Peptides

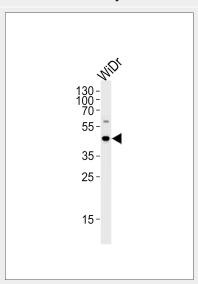


- Dot Blot
- Immunohistochemistry
- <u>Immunofluorescence</u>
- <u>Immunoprecipitation</u>
- Flow Cytomety
- Cell Culture

C9orf72 Antibody (C-term) - Images



C9orf72 Antibody (C-term) (Cat. #AP12928b) western blot analysis in MDA-MB231 cell line lysates (35ug/lane). This demonstrates the C9orf72 antibody detected the C9orf72 protein (arrow).



C9orf72 Antibody (C-term) (Cat.# AP12928b) western blot analysis in WiDr cell lysate (35ug/lane). This demonstrates that the C9orf72 antibody detected C9orf72 protein (arrow).

C9orf72 Antibody (C-term) - 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)

C9orf72 Antibody (C-term) - Citations

• Novel antibodies reveal presynaptic localization of C9orf72 protein and reduced protein levels in C9orf72 mutation carriers.





• Immunohistochemical detection of C9orf72 protein in frontotemporal lobar degeneration and motor neurone disease: patterns of immunostaining and an evaluation of commercial antibodies.

- Loss-of-function mutations in the C9ORF72 mouse ortholog cause fatal autoimmune disease.
- The ALS/FTLD associated protein C9orf72 associates with SMCR8 and WDR41 to regulate the autophagy-lysosome pathway.