

### C9orf72 Antibody (C-term)

Affinity Purified Rabbit Polyclonal Antibody (Pab) Catalog # AP12928b

#### Specification

# C9orf72 Antibody (C-term) - Product Information

Application Primary Accession Other Accession Reactivity Predicted Host Clonality Isotype Calculated MW Antigen Region WB,E <u>O96LT7</u> <u>O66HC3, NP\_060795.1, NP\_659442.2, O6DFW0</u> Human Mouse, Rat Rabbit Polyclonal Rabbit IgG 54328 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 (<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:<u>27103069</u>,

PubMed:27193190, PubMed:27617292, PubMed:28195531, PubMed:37821429). Acts as a GEF for RAB39A which enables HOPS-mediated autophagosome-lysosome membrane tethering and fusion in mammalian autophagy (PubMed:<u>37821429</u>). 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:000250|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

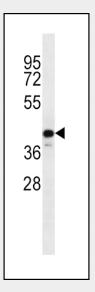
Provided below are standard protocols that you may find useful for product applications.

- <u>Western Blot</u>
- Blocking Peptides

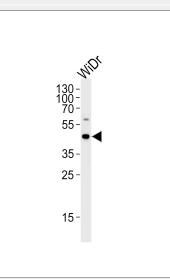


- Dot Blot
- Immunohistochemistry
- Immunofluorescence
- Immunoprecipitation
- Flow Cytomety
- <u>Cell Culture</u>

### 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.