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C9orf72 Antibody (C-term)

Affinity Purified Rabbit Polyclonal Antibody (Pab)
Catalog # AP12928b

Product Information

Application	WB, E
Primary Accession	Q96LT7
Other Accession	Q66HC3, NP_060795.1, NP_659442.2
Reactivity	Human, Rat, Mouse
Predicted	Rat
Host	Rabbit
Clonality	Polyclonal
Isotype	Rabbit IgG
Calculated MW	54328
Antigen Region	396-424

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.

Protein Information

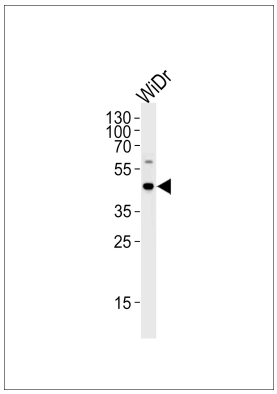
Name	C9orf72 (HGNC:28337)
Function	Acts as a guanine-nucleotide releasing factor (GEF) for Rab GTPases by promoting the conversion of inactive RAB-GDP to the active form RAB-GTP (PubMed:27103069, PubMed:27193190, PubMed:27617292, PubMed:28195531, PubMed:37821429). Acts as a GEF for RAB39A which

	enables HOPS-mediated autophagosome-lysosome membrane tethering and fusion in mammalian autophagy (PubMed: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).

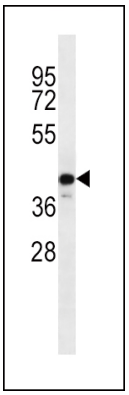
References

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)

Images



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

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.