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Anti-C9orf72 Antibody

Catalog # AP95712

Product Information

Application	WB, IHC, FC, IF/IC
Primary Accession	Q96LT7
Reactivity	Human, Mouse, Rat, Mk
Host	Mouse
Clonality	Monoclonal
Calculated MW	54328

Additional Information

Gene ID	203228
Other Names	Protein C9orf72
Target/Specificity	Recombinant fusion protein of human C9orf72 expressed in E. Coli
Dilution	WB~~WB (1/500 - 1/1000) IHC~~1:100~500 FC~~FC (1/100 - 1/200) IF/IC~~N/A
Format	Mouse IgG2b. Liquid in PBS, pH 7.3, 30% glycerol, and 0.01% sodium azide.
Storage	Store at -20 °C.Stable for 12 months from date of receipt

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).

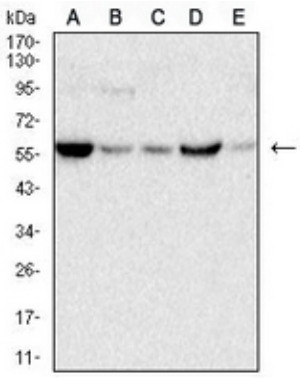
Cellular Location

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). Primarily localized in cytosol under basal conditions (PubMed:37821429). Primarily gathered on autolysosome 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

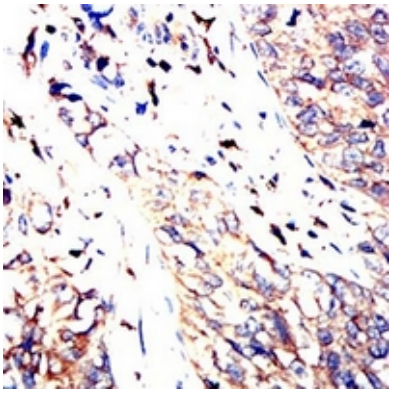
Both isoforms are widely expressed, including kidney, lung, liver, heart, testis and several brain regions, such as cerebellum. Also expressed in the frontal cortex and in lymphoblasts (at protein level).

Images

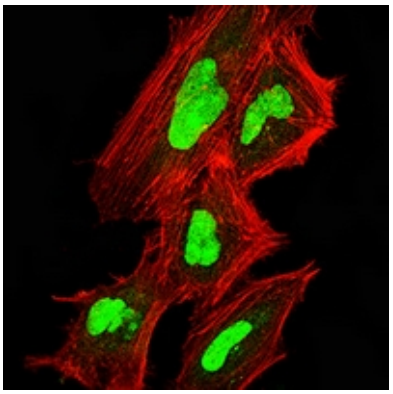


Western blot analysis of C9orf72 expression in C6 (A), PC12 (B), COS7 (C), NIH/3T3 (D), SKNSH (E) whole cell lysates. (Predicted band size: 54 kD; Observed band size: 56 kD)

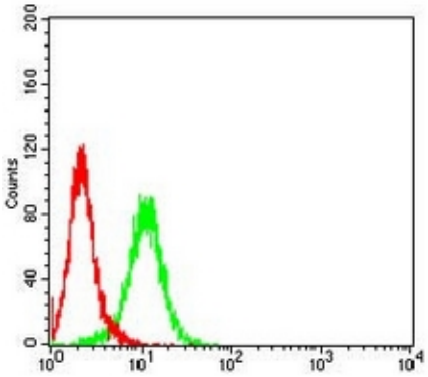
Immunohistochemical analysis of C9orf72 staining in human bladder cancer formalin fixed paraffin embedded tissue section. The section was pre-treated using heat mediated antigen retrieval with sodium citrate buffer (pH



6.0). The section was then incubated with the antibody at room temperature and detected using an HRP conjugated compact polymer system. DAB was used as the chromogen. The section was then counterstained with haematoxylin and mounted with DPX.



Immunofluorescent analysis of C9orf72 staining in Hela cells. Formalin-fixed cells were permeabilized with 0.1% Triton X-100 in TBS for 5-10 minutes and blocked with 3% BSA-PBS for 30 minutes at room temperature. Cells were probed with the primary antibody in 3% BSA-PBS and incubated overnight at 4 °C in a hidified chamber. Cells were washed with PBST and incubated with an AF488-conjugated secondary antibody (green) in PBS at room temperature in the dark. Phalloidin - AF594 was used to stain Actin filaments (red). DAPI was used to stain the cell nuclei (blue).



Flow cytometric analysis of HeLa cells using Anti-C9orf72 Antibody (green) and negative control (red).

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