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ALSFTD Antibody

Catalog # ASC11967

Product Information

Application	WB, ICC, E
Primary Accession	Q96LT7
Other Accession	NP_060795, 37039612
Reactivity	Human, Mouse, Rat
Host	Rabbit
Clonality	Polyclonal
Isotype	IgG
Calculated MW	54328
Concentration (mg/ml)	1 mg/mL
Conjugate	Unconjugated
Application Notes	ALSFTD antibody can be used for the detection of ALSFTD by Western blot at 1 - 2 □g/mL. Antibody can also be used for immunocytochemistry at 10 □g/ml.

Additional Information

Gene ID	203228
Other Names	Protein C9orf72, C9orf72
Target/Specificity	ALSFTD; ALSFTD antibody is human, mouse and rat reactive. At least two isoforms are known to exist.
Reconstitution & Storage	ALSFTD antibody can be stored at 4℃ for three months and -20℃, stable for up to one year.
Precautions	ALSFTD Antibody 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). 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).

Background

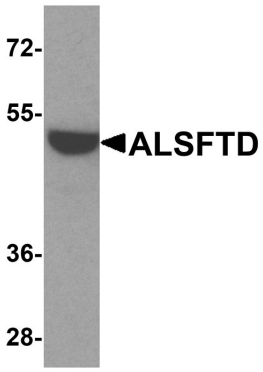
ALSFTD (C9orf72) is considered to play a role in gender determination (1). Hereditary hemorrhagic telangiectasia, which is characterized by harmful vascular defects, is associated with the chromosome 9 gene encoding endoglin protein, ENG (2). Familial dysautonomia is also associated with chromosome 9 though through the gene IKBKAP. Notably, chromosome 9 encompasses the largest interferon family gene cluster (3,4).

References

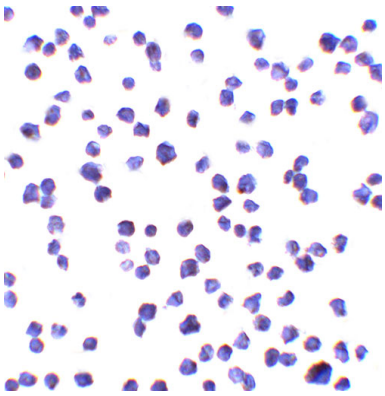
Takada LT and Sha SJ. Neuropsychiatric features of C9orf72-associated behavioral variant frontotemporal dementia and frontotemporal dementia with motor neuron disease. Alzheimers Res. Ther. 2012; 4:38.
Coon EA, Whitwell JL, Parisi JE, et al. Right temporal variant frontotemporal dementia with motor neuron

disease. J. Clin. Neurosci. 2012; 19:85-91.
Snowden JS, Rollinson S, Thompson JC, et al. Distinct clinical and pathological characteristics of frontotemporal dementia associated with C9ORF72 mutations. Brain 2012; 135:693-708.
Wen X, Tan W, Westergard T, et al. Antisense proline-arginine RAN dipeptides linked to C9ORF72-ALS/FTD form toxic nuclear aggregates that initiate in vitro and in vivo neuronal death. Neuron 2014; 84:1213-25.

Images



Western blot analysis of ALSFTD in HeLa cell lysate with ALSFTD antibody at 1 µg/ml.



Immunocytochemistry of ALSFTD in A-20 cells with ALSFTD antibody at 10 µg/ml.

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