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ALSFTD Antibody
Catalog # ASC11967

Specification

ALSFTD Antibody - Product Information

Application	**WB, ICC**
Primary Accession	Q96LT7
Other Accession	NP_060795, 37039612
Reactivity	**Human, Mouse, Rat**
Host	**Rabbit**
Clonality	**Polyclonal**
Isotype	**IgG**
Calculated MW	**Predicted: 53 kDa** **Observed: 52 kDa KDa**
Application Notes	**ALSFTD antibody can be used for the detection of ALSFTD by Western blot at 1 - 2 µg/mL. Antibody can also be used for immunocytochemistry at 10 µg/ml.**

ALSFTD Antibody - Additional Information

Gene ID **203228**

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.

ALSFTD Antibody - Protein Information

Name C9orf72 (HGNC:28337)

Function
Component of the C9orf72-SMCR8 complex, a complex that has guanine nucleotide exchange factor (GEF) activity and regulates autophagy (PubMed:27103069, PubMed:27193190, PubMed:27617292, PubMed:28195531, PubMed:32303654). In the complex, C9orf72 and SMCR8 probably constitute the catalytic subunits that promote the exchange of GDP to GTP, converting inactive GDP-bound RAB8A and RAB39B into their active GTP-bound form, thereby promoting autophagosome maturation (PubMed:<a

href="http://www.uniprot.org/citations/27103069" target="_blank">27103069). 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). As part of the C9orf72-SMCR8 complex, stimulates RAB8A and RAB11A GTPase activity in vitro (PubMed:32303654). Positively regulates 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
Nucleus. Cytoplasm. Cytoplasm, P-body. Cytoplasm, Stress granule. Endosome Lysosome Cytoplasmic vesicle, autophagosome 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). {ECO:0000250|UniProtKB:Q6DFW0, ECO:0000269|PubMed:21944778, ECO:0000269|PubMed:21944779, ECO:0000269|PubMed:27037575} [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).

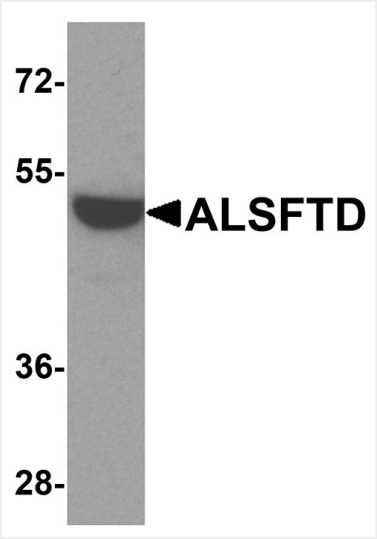
ALSFTD Antibody - Protocols

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

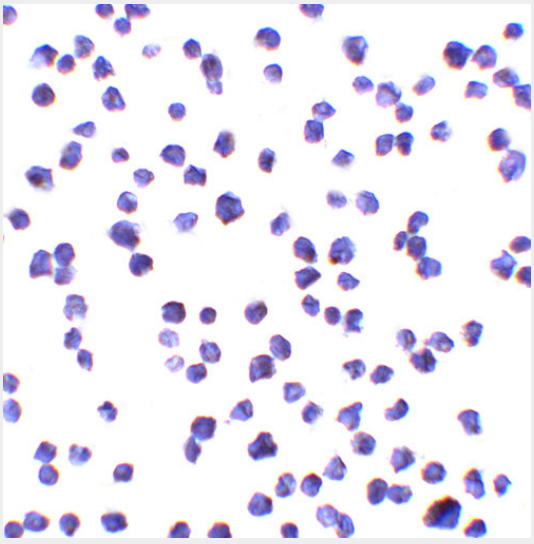
- Western Blot
- Blocking Peptides
- Dot Blot
- Immunohistochemistry
- Immunofluorescence
- Immunoprecipitation

- Flow Cytomety
- Cell Culture

ALSFTD Antibody - 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.

ALSFTD Antibody - 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).

ALSFTD Antibody - 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.