Revision 1

Store at -20C

#64196

C9orf72 Antibody

Cell Signaling TECHNOLOGY®

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For Research Use Only. Not for Use in Diagnostic Procedures.

Applications:	Reactivity:	Sensitivity:	MW (kDa):	Source/Isotype:	UniProt ID:	Entrez-Gene Id:
W	H M R	Endogenous	49	Rabbit	#Q96LT7	203228

Product Usage Information

Application	Dilution
Western Blotting	1:1000

Storage

Supplied in 10 mM sodium HEPES (pH 7.5), 150 mM NaCl, 100 µg/ml BSA and 50% glycerol. Store at –20°C. *Do not aliquot the antibody.*

Specificity/Sensitivity

C9orf72 Antibody recognizes endogenous levels of total C9orf72 protein.

Source / Purification

Polyclonal antibodies are produced by immunizing animals with a synthetic peptide corresponding to residues surrounding Ala202 of human C9orf72 protein. Antibodies are purified by protein A and peptide affinity chromatography.

Background

The expansion of hexanucleotide GGGGCC repeats in the *C9orf72* gene causes chromosome 9p-linked neurodegenerative diseases amyotrophic lateral sclerosis (ALS) and frontotemporal dementia (FTD) (1,2). The specific mechanism by which of these repeats contributes to disease etiology is currently an active area of investigation (3). Several gain of function mechanisms have been proposed. These mechanisms include toxicity from *C9orf72* RNA containing the hexanucleotide repeats (4) and toxicity generated from dipeptide repeat proteins produced by repeat-associated non-ATG translation (5). In addition to gain of function mechanisms, the genetic hexanucleotide repeat expansions may cause a loss of function of the C9orf72 protein. C9orf72 contains a predicted DENN (differentially expressed in normal and neoplastic cells) domain that typically functions as guanine exchange factors for Rab GTPases, proteins that play key regulatory roles in membrane trafficking (6). Consistent with C9orf72 normally functioning in membrane trafficking, biochemical and genetic studies revealed that C9orf72 forms a protein complex with Sim-Magenis chromosome region 8 (SMCR8) and WD repeat-containing protein 41 (WDR41) to regulate the autophagy-lysosomal pathway (7), suggesting that C9orf72-dependent alterations in the autophagy-lysosomal pathway might contribute to ALS/FTD pathology.

Background References

1. DeJesus-Hernandez, M. et al. (2011) *Neuron* 72, 245-56.
2. Renton, A.E. et al. (2011) *Neuron* 72, 257-68.
3. Balendra, R. and Isaacs, A.M. (2018) *Nat Rev Neurol* 14, 544-58.
4. Zhang, Y.J. et al. (2019) *Science* 363, pii: eaav2606. doi: 10.1126/science.aav2606.
5. Cleary, J.D. et al. (2018) *J Biol Chem* 293, 16127-41.
6. Amick, J. and Ferguson, S.M. (2017) *Traffic* 18, 267-76.
7. Sullivan, P.M. et al. (2016) *Acta Neuropathol Commun* 4, 51.

Species Reactivity

Species reactivity is determined by testing in at least one approved application (e.g., western blot).

Western Blot Buffer

IMPORTANT: For western blots, incubate membrane with diluted primary antibody in 5% w/v BSA, 1X TBS, 0.1% Tween® 20 at 4°C with gentle shaking, overnight.

Applications Key

W: Western Blotting

Cross-Reactivity Key

H: Human **M:** Mouse **R:** Rat

Trademarks and Patents

Limited Uses

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