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C9orf72

Reactivity: Human

Tested applications:WB

Recommended Dilution:WB 1:200 - 1:1000

Calculated MW:54kDa

Observed MW: Refer to figures

Immunogen:

Recombinant protein of human C9orf72

Storage Buffer:

Store at 4. Avoid freeze / thaw cycles. Buffer: PBS with 0.02% sodium azide, 50% glycerol, pH7.3.

Synonym:

ALSFTD; FTDALS

Catalog #:A9532

Antibody Type:

Polyclonal Antibody

Species: Rabbit

Gene ID:203228

Isotype: IgG

Swiss Prot:Q96LT7

Purity: Affinity purification

For research use only.

Background:

The protein encoded by this gene plays an important role in the regulation of endosomal trafficking, and has been shown to interact with Rab proteins that are involved in autophagy and endocytic transport. Expansion of a hexanucleotide repeat in non-coding sequence between alternate 5' exons in transcripts from this gene is associated with 9p-linked ALS (amyotrophic lateral sclerosis) and FTD (frontotemporal dementia) (PMID: 21944778, 21944779). Studies suggest that hexanucleotide expansions could result in the selective stabilization of repeat-containing pre-mRNA, and the accumulation of insoluble dipeptide repeat protein aggregates that could be pathogenic in FTD-ALS patients (PMID: 23393093). Alternative splicing results in multiple transcript variants encoding different isoforms.

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