

## Recombinant Human C9orf72 Protein, GST-Tagged

**Cat. No.** C9orf72-0211H    **Lot. No.** (See product label)

### SPECIFICATION

<b>Product Overview</b>	Human C9orf72 full-length ORF (NP_060795.1, 1 a.a. - 481 a.a.) recombinant protein with GST tag at N-terminal.
<b>Species</b>	Human
<b>Source</b>	Wheat Germ
<b>Description</b>	<p>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 GGGGCC repeat from 2-22 copies to 700-1600 copies in the intronic 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. [provided by RefSeq, Jul 2016]</p>
<b>Molecular Mass</b>	80.7 kDa
<b>AA Sequence</b>	<p>MSTLCPPPPSPAVAKTEIALSGKSPLLAATFAYWDNILGPRVRHIWAPKTEQVLLSDGE ITFLANHTLNGEILRNAESGAIDVKFFVLSEKGVIIIVSLIFDGNWNGDRSTYGLSIIIPQT ELSFYLP LHRVCVDRLTHIIRKGR IWMHKERQENVQKIILEGTERMEDQGQSIIPMLT</p>

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GEVIPVMELLSSMKSHSVPEEIDIADTVLNDDDIGDSCHEGFLNNAISSHLQTCGCSV  
 VVGSSAEKVNKIVRTLCLFLTPAERKCSRLCEAESSFKYESGLFVQGLLKDSTGGSFVL  
 PFRQVMYAPYPTTHIDVDVNTVKQMPPCHEHIYNQRRYMRSELTAFWRATSEEDM  
 AQDTIITYTDESFTPDNLIFQDVLHRDTLVKAFLDQVFQLKPGLSLRSTFLAQFLLVLHR  
 KALTLIKYIEDDTQKGKKPFKSLRNLIKIDLDLTAEGDLNIIMALAEKIKPGLHSFIFGRPF  
 YTSVQERDVLMTF

<b>Applications</b>	Enzyme-linked Immunoabsorbent Assay Western Blot (Recombinant protein) Antibody Production Protein Array
<b>Notes</b>	Best use within three months from the date of receipt of this protein.
<b>Storage</b>	Store at -80 centigrade. Aliquot to avoid repeated freezing and thawing.
<b>Storage Buffer</b>	50 mM Tris-HCl, 10 mM reduced Glutathione, pH=8.0 in the elution buffer.

## GENE INFORMATION

<b>Gene Name</b>	C9orf72 chromosome 9 open reading frame 72 [ Homo sapiens (human) ]
<b>Official Symbol</b>	C9orf72
<b>Synonyms</b>	C9orf72; chromosome 9 open reading frame 72; ALSFTD; FTDALS; DENNL72; FTDALS1; guanine nucleotide exchange C9orf72; protein C9orf72; Chromosome 9 Open Reading Frame 72; Guanine Nucleotide Exchange C9orf72; Protein C9orf72; DENNL72; FTDALS1; ALSFTD; FTDALS
<b>Gene ID</b>	203228

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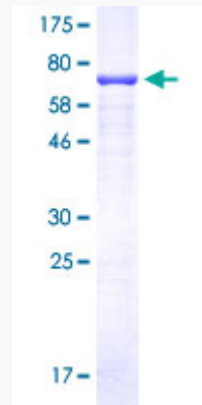
mRNA Refseq [NM\\_001256054](#)

Protein Refseq [NP\\_001242983](#)

MIM [614260](#)

UniProt ID [Q96LT7](#)

**Quality Control  
Testing**



12.5% SDS-PAGE Stained with Coomassie Blue

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