

Recombinant Full Length Human C9orf72 Protein, GST-tagged

Cat. No. C9orf72-2722HF **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Human C9orf72 full-length ORF (NP_060795.1, 1 a.a. - 481 a.a.) recombinant protein with GST tag at N-terminal.
Species	Human
Source	In Vitro Cell Free System
ProteinLength	481 amino acids
Description	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]
Molecular Mass	80.7 kDa
AA Sequence	MSTLCPPPPSPAVAKTEIALSGKSPLLAATFAYWDNILGPRVRHIWAPKTEQVLLSDGE ITFLANHTLNGEILRNAESGAIDVKFFVLSEKGVIIIVSLIFDGNWNGDRSTYGLSILPQT

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ELSFYLPLHRVCVDRLTHIIRKGRIMHKERQENVQKIILEGTERMEDQGQSIIPMLT
 GEVIPVMELLSSMKSHSVPEEIDIADTVLNDDDIGDSCHEGFLNNAISSHLQTCGCSV
 VVGSSAEKVNKIVRTLCLFLTPAERKCSRCEAESSFKYESGLFVQGLLKDSTGSLV
 PFRQVMYAPYPTTHIDVDVNTVKQMPPCHEHIYNQRRYMRSELTAFWRATSEEDM
 AQDTIIYTDESFTPDLNIFQDVLHRDTLVKAFLDQVFQLKPGLSLRSTFLAQFLLVLHR
 KALTLIKYIEDDTQKGKKPFKSLRNLKIDDLTAEGDLNIIMALAEKIKPGLHSFIFGRPF
 YTSVQERDVLMTF

Applications

Enzyme-linked Immunoabsorbent Assay
 Western Blot (Recombinant protein)
 Antibody Production
 Protein Array

Notes

Best use within three months from the date of receipt of this protein.

Storage

Store at -80 centigrade. Aliquot to avoid repeated freezing and thawing.

Storage Buffer

50 mM Tris-HCl, 10 mM reduced Glutathione, pH=8.0 in the elution buffer.

GENE INFORMATION

Gene Name

C9orf72 chromosome 9 open reading frame 72 [Homo sapiens (human)]

Official Symbol

C9orf72

Synonyms

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

Gene ID

203228

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mRNA Refseq	NM_001256054
Protein Refseq	NP_001242983
MIM	614260
UniProt ID	Q96LT7

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