

Recombinant Human C9orf72 Protein, His-tagged

Cat. No. C9orf72-2037H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant human C9orf72 protein, fused to His-tagged at N-terminus, was expressed in E. coli
Species	Human
Source	E.coli
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].
Form	25 mM Tris, pH 8.0, 500mM NaCl, 10% glycerol.
Molecular Mass	24.6 kDa
Purity	> 80% as determined by SDS-PAGE and Coomassie blue staining

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Concentration >50 ug/mL as determined by microplate BCA method

GENE INFORMATION

Gene Name C9orf72 chromosome 9 open reading frame 72 [Homo sapiens]

Official Symbol C9orf72

Synonyms ALSFTD; DENNL72; FTDALS; FTDALS1

Gene ID 203228

mRNA Refseq NM_145005.6

Protein Refseq NP_659442

MIM 614260

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