

Product datasheet for **TP761492**

C9orf72 (NM_018325) Human Recombinant Protein

Product data:

| | |
|---------------------------------------|---|
| Product Type: | Recombinant Proteins |
| Description: | Purified recombinant protein of Human chromosome 9 open reading frame 72 (C9orf72), transcript variant 2, full length, with N-terminal HIS tag, expressed in E.coli, 50ug |
| Species: | Human |
| Expression Host: | E. coli |
| Expression cDNA Clone or AA Sequence: | A DNA sequence encoding human full-length C9orf72 |
| Tag: | N-His |
| Predicted MW: | 54.1 kDa |
| Concentration: | >0.05 µg/µL as determined by microplate BCA method |
| Purity: | > 80% as determined by SDS-PAGE and Coomassie blue staining |
| Buffer: | 25 mM Tris-HCl, pH 8.0, 150 mM NaCl, 1% sarkosyl, 10% glycerol |
| Note: | For testing in cell culture applications, please filter before use. Note that you may experience some loss of protein during the filtration process. |
| Storage: | Store at -80°C. |
| Stability: | Stable for 12 months from the date of receipt of the product under proper storage and handling conditions. Avoid repeated freeze-thaw cycles. |
| RefSeq: | NP_060795 |
| Locus ID: | 203228 |
| UniProt ID: | Q96LT7 |
| RefSeq Size: | 3244 |
| Cytogenetics: | 9p21.2 |
| RefSeq ORF: | 1443 |
| Synonyms: | ALSFTD; DENND9; DENNL72; FTDALS; FTDALS1 |



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Summary:

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]

Product images: