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Product datasheet for TP760931

C9orf72 (NM_145005) Human Recombinant Protein

Product data:

Product Type:	Recombinant Proteins
Description:	Purified recombinant protein of Human chromosome 9 open reading frame 72 (C9orf72), transcript variant 1, 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:	24.6 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, 500 mM NaCl, 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_659442
Locus ID:	203228
UniProt ID:	Q96LT7
RefSeq Size:	1882
Cytogenetics:	9p21.2
RefSeq ORF:	666
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: