

Recombinant Full Length Human C9orf72 Protein, N-His-tagged

Cat. No. C9orf72-678HFL **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant Full Length Human C9orf72 Protein, fused to His-tag 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). 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. Alternative splicing results in multiple transcript variants encoding different isoforms.
Form	25mM Tris, pH8.0, 150mM NaCl, 10% glycerol, 1% Sarkosyl.
Molecular Mass	54.1 kDa
AA Sequence	A DNA sequence encoding human full-length C9orf72.

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Purity	> 80% as determined by SDS-PAGE and Coomassie blue staining.
Stability	Stable for 12 months from the date of receipt of the product under proper storage and handling conditions. Avoid repeated freeze-thaw cycles.
Storage	Store at -80 centigrade.
Concentration	>50 ug/mL as determined by microplate BCA method.
Full Length	Full L.

GENE INFORMATION

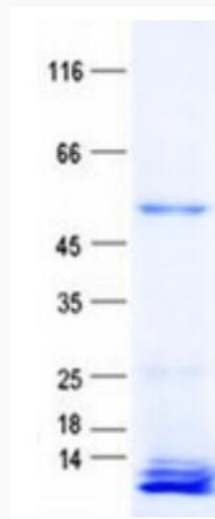
Gene Name	C9orf72 C9orf72-SMCR8 complex subunit [Homo sapiens (human)]
Official Symbol	C9orf72
Synonyms	ALSFTD; DENND9; FTDALS; DENNL72; FTDALS1
Gene ID	203228
mRNA Refseq	NM_018325.5
Protein Refseq	NP_060795.1
MIM	614260
UniProt ID	Q96LT7

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SDS-PAGE



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