

Recombinant Human ATXN2 Protein, His-Tagged

Cat. No. ATXN2-01H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant Human ATXN2 Protein, His-tagged was expressed in E.coli cell
Species	Human
Source	E.coli
ProteinLength	701-1180 aa
Description	This gene belongs to a group of genes that is associated with microsatellite-expansion diseases, a class of neurological and neuromuscular disorders caused by expansion of short stretches of repetitive DNA. The protein encoded by this gene has two globular domains near the N-terminus, one of which contains a clathrin-mediated trans-Golgi signal and an endoplasmic reticulum exit signal. The encoded cytoplasmic protein localizes to the endoplasmic reticulum and plasma membrane, is involved in endocytosis, and modulates mTOR signals, modifying ribosomal translation and mitochondrial function. The N-terminal region of the protein contains a polyglutamine tract of 14-31 residues that can be expanded in the pathogenic state to 32-200 residues. Intermediate length expansions of this tract increase susceptibility to amyotrophic lateral sclerosis, while long expansions of this tract result in spinocerebellar ataxia-2, an autosomal-dominantly inherited, neurodegenerative disorder. Genome-wide association studies indicate that loss-of-function mutations in this gene may be associated with susceptibility to type I diabetes, obesity and hypertension. Alternative splicing results in multiple transcript variants.

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Molecular Mass	The protein has a calculated MW of 53.13 kDa.
AA Sequence	MTPSGPVLASPQAGIIPTEAVAMPIPAASPTPASPASNRAVTPSSEAKDSRLQDQRQ NSPAGNKENIKPNETSPSFSKAENKGISPVVSEHRKQIDDLKKFKNDFRLQPSSTSE SMDQLLNKNREGEKSRDLIKDKIEPSAKDSFIENSSSNCTSGSSKPNSPSISPSILSNT EHKRGPEVTSQGVQTSSPACKQEKKDDKEEKKDAAEQVRKSTLNPNAKEFNPRSFS QPKPSTTPTSPRPQAQPSPSMVGHQQPTPVYTQPVCFAPNMMYPVPVSPGVQPLY PIPMTPMPVNQAKTYRAVPNMPQQRQDQHHQSAMMHPASAAGPPIAATPPAYSTQ YVAYSPQQFPNQPLVQHVPHYQSQHHPHVYSPVIQGNARMMAPPTHAQPGLVSSSA TQYGAHEQTHAMYACPKLPYNKETSPSFYFAISTGSLAQQYAHNPATLHPHTPHPQ PSATPTGQQSQHGGSHPPAPSPVQHHQHQAQLEHHHHHH
Purity	80% by SDS-PAGE
Storage	Store it at -20 to -80 °C. It is recommended that the protein be aliquoted for optimal storage. Avoid repeated freeze-thaw cycles.
Concentration	0.5 mg/mL
Storage Buffer	PBS, 300mM NaCl, 10% glycerol, pH 7.4
GENE INFORMATION	
Gene Name	ATXN2 ataxin 2 [Homo sapiens (human)]
Official Symbol	ATXN2
Synonyms	ATX2; SCA2; TNRC13
Gene ID	6311

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mRNA Refseq	NM_001310121.1
Protein Refseq	NP_001297050.1
MIM	601517
UniProt ID	F8VQP2

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