

Recombinant Human ATXN2 Protein, GST-tagged

Cat. No. ATXN2-156H **Lot. No.** (See product label)

SPECIFICATION

Product Overview

Recombinant Human ATXN2 Protein(NP_001297050.1)(251-600 aa), fused with N-terminal GST tag, was expressed in E. coli.

Species

Human

Source

E.coli

ProteinLength

251-600 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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Form	The purified protein was Lyophilized from sterile PBS (58mM Na ₂ HPO ₄ , 17mM NaH ₂ PO ₄ , 68mM NaCl, pH8.). 5 % trehalose and 5 % mannitol are added as protectants before lyophilization.
Bio-activity	Not tested.
AA Sequence	GSDQRVVNGGVPWPSPCSPSSRPPSRYPQSGPNSLPPRAATPTRPPSRPPSRPS RPPSHPSAHGSPAPVSTMPKRMSSSEGPSPRMSPKAQRHPRNHRVSAGRGSISSGLE FVSHNPPSEAAATPPVARTSPSGGTWSSVVSQVPRLSPKTHRPRSPRQNSIGNTPSG PVLASPQAGIIPTEAVAMPIAASPTPASPASNRAVTPSSEAKDSRLQDQRQNSPAG NKENIKPNETSPSFSKAENKGISPVVSEHRKQIDDLKKFKNDFRLQPSSTSESMDQL LNKNREGEKSRDLIKDKIEPSAKDSFIENSSSNTSGSSKPNSPSISPSILSNTTEHKRG PEVTSQGVQTSS
Purity	75%, by SDS-PAGE with Coomassie Brilliant Blue staining.
Applications	Blocking peptide
Stability	Store for up to 12 months at -20°C to -80°C as lyophilized powder.
Storage	Short-term storage: Store at 2-8°C for (1-2 weeks). Long-term storage: Aliquot and store at -20°C to -80°C for up to 3 months, buffer containing 50% glycerol is recommended for reconstitution. Avoid repeat freeze-thaw cycles.
Reconstitution	Reconstitute at 0.25 g/μl in 200 μl sterile water for short-term storage. Reconstitution with 200 μl 50% glycerol solution is recommended for longer term storage (see Stability and Storage for more details). If a different concentration is needed for your purposes please adjust the reconstitution volume as required (please note: the ion concentration of the final

solution will vary according to the volume used).

Note: Centrifuge vial before opening. When reconstituting, gently pipet and wash down the sides of the vial to ensure full recovery of the protein into solution.

Shipping

The product is shipped at ambient temperature. Upon receipt, store it immediately at the recommended temperature (see below).

GENE INFORMATION

Gene Name ATXN2 ataxin 2 [Homo sapiens (human)]

Official Symbol ATXN2

Synonyms ATX2; SCA2; TNRC13

Gene ID 6311

mRNA Refseq NM_001310121.1

Protein Refseq NP_001297050.1

MIM 601517

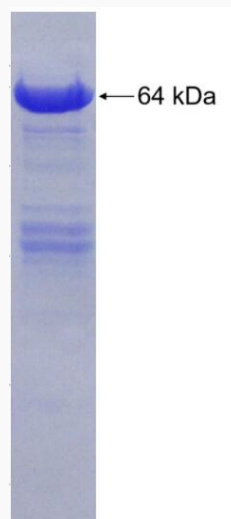
UniProt ID Q99700

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**SDS-PAGE of
ATXN2-156H**



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