

Recombinant Human ATXN2 Protein, Non tagged

Cat. No. ATXN2-10069H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant Human ATXN2 Protein without tag was expressed in E. coli.
Species	Human
Source	E.coli
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.
Molecular Mass	37 kDa

 Tel: 1-631-559-9269 1-516-512-3133

 Email: info@creative-biomart.com  Fax: 1-631-938-8127

 45-1 Ramsey Road, Shirley, NY 11967, USA

AA Sequence	MGSDQRVVNGGVPWSPCSPSSRPPSRYPQSGPNSLPPRAATPTRPPSRPPSRP SRPPSHPSAHGSPAPVSTMPKRMSSEGPPRMSPKAQRHPRNHRVSAGRGSISSGL EFVSHNPPSEAATPPVARTSPSGGTWSSVSGVPRLSPKTHRPRSPRQNSIGNTPS GPVLASPQAGIIPTEAVAMPIPAASPTPASPASNRAVTPSSEAKDSRLQDQRQNSPA GNKENIKPNETSPSFSKAENKGISPVVSEHRKQIDDLKKFKNDFRLQPSSTSESMDQ LLNKNREGEKSRDLIKDIEPSAKDSFIENSSSNCTSGSSKPNSPSISPSILSNTTEHKR GPEVTSQGVQTSS
Purity	> 90% by SDS-PAGE
Storage	Store it under sterile conditions at -20 to -80 centigrade. It is recommended that the protein be aliquoted for optimal storage. Avoid repeated freeze-thaw cycles.
Concentration	0.17 mg/mL
Storage Buffer	Sterile 50mM Tris, 300mM NaCl, 10% Glycerol, pH7.5
GENE INFORMATION	
Gene Name	ATXN2 ataxin 2 [Homo sapiens (human)]
Official Symbol	ATXN2
Synonyms	ATXN2; ataxin 2; SCA2, spinocerebellar ataxia 2 (olivopontocerebellar ataxia 2, autosomal dominant, ataxin 2), TNRC13; ataxin-2; ATX2; trinucleotide repeat containing 13; spinocerebellar ataxia type 2 protein; trinucleotide repeat-containing gene 13 protein; SCA2; TNRC13; FLJ46772;
Gene ID	6311
mRNA Refseq	NM_002973

 Tel: 1-631-559-9269 1-516-512-3133

 Email: info@creative-biomart.com  Fax: 1-631-938-8127

 45-1 Ramsey Road, Shirley, NY 11967, USA

Protein Refseq	NP_002964
MIM	601517
UniProt ID	Q99700

 Tel: 1-631-559-9269 1-516-512-3133

 Email: info@creative-biomart.com  Fax: 1-631-938-8127

 45-1 Ramsey Road, Shirley, NY 11967, USA