

Recombinant Human ATXN2 protein, GST-tagged

Cat. No. ATXN2-1031H Lot. No. (See product label)

SPECIFICATION

Product Overview	Human ATXN2 partial ORF (NP_002964, 1214 a.a. - 1313 a.a.) recombinant protein with GST-tag at N-terminal.
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Species	Human
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Source	Wheat Germ
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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. [provided by RefSeq, Nov 2016]

 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

Molecular Mass	36.74 kDa
AA Sequence	PQNSFPAAQQTVFTIHPSHVQPAYTNPPHMAHVPQAHVQSGMVPSHPTAHAPMML MTTQPPGGPQAALASALQPIPVSTTAHFPYMTGPSVQAHHQQQL
Applications	Enzyme-linked Immunoabsorbent Assay; Western Blot (Recombinant protein); Antibody Production; Protein Array
Notes	Best use within three months from the date of receipt of this protein.
Storage	Store at -80 centigrade. Aliquot to avoid repeated freezing and thawing.
Storage Buffer	50 mM Tris-HCl, 10 mM reduced Glutathione, pH=8.0 in the elution buffer.

GENE INFORMATION

Gene Name	ATXN2 ataxin 2 [Homo sapiens]
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
Protein Refseq	NP_002964

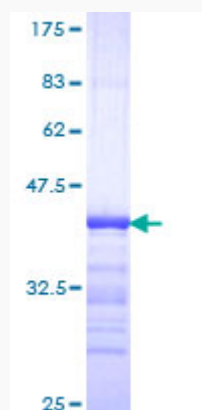
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MIM 601517

UniProt ID Q99700



12.5% SDS-PAGE Stained with Coomassie Blue.

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