Recombinant Human ATXN1 protein, GST-tagged

Cat. No. ATXN1-1029H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Human ATXN1 partial ORF (NP_000323, 576 a.a. - 675 a.a.) recombinant protein with GST-tag at N-terminal.
Species	Human
Source	Wheat Germ
Description	The autosomal dominant cerebellar ataxias (ADCA) are a heterogeneous group of neurodegenerative disorders characterized by progressive degeneration of the cerebellum, brain stem and spinal cord. Clinically, ADCA has been divided into three groups: ADCA types I-III. ADCAI is genetically heterogeneous, with five genetic loci, designated spinocerebellar ataxia (SCA) 1, 2, 3, 4 and 6, being assigned to five different chromosomes. ADCAII, which always presents with retinal degeneration (SCA7), and ADCAIII often referred to as the `pure' cerebellar syndrome (SCA5), are most likely homogeneous disorders. Several SCA genes have been cloned and shown to contain CAG repeats in their coding regions. ADCA is caused by the expansion of the CAG repeats, producing an elongated polyglutamine tract in the corresponding protein. The expanded repeats are variable in size and unstable, usually increasing in size when transmitted to successive generations. The function of the ataxins is not known. This locus has been mapped to chromosome 6, and it has been determined that the diseased allele contains 40-83 CAG repeats, compared to 6-39 in the normal allele, and is associated with spinocerebellar ataxia type 1 (SCA1). At least two transcript variants encoding the same protein have been found for this gene. [provided by RefSeq, Jul 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	KGSIIQLANGELKKVEDLKTEDFIQSAEISNDLKIDSSTVERIEDSHSPGVAVIQFAVGE HRAQVSVEVLVEYPFFVFGQGWSSCCPERTSQLFDLPCSK
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	ATXN1 ataxin 1 [Homo sapiens]
Official Symbol	ATXN1
Synonyms	ATXN1; ataxin 1; SCA1, spinocerebellar ataxia 1 (olivopontocerebellar ataxia 1, autosomal dominant, ataxin 1); ataxin-1; ATX1; D6S504E; spinocerebellar ataxia type 1 protein; SCA1;
Gene ID	6310
mRNA Refseq	NM_000332
Protein Refseq	NP_000323
MIM	601556

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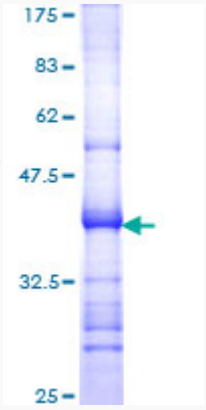
UniProt ID	P54253



12.5% SDS-PAGE Stained with Coomassie Blue.

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