

Recombinant Full Length Human ATXN1 Protein, C-Flag-tagged

Cat. No. ATXN1-372HFL **Lot. No.** (See product label)

SPECIFICATION

Product Overview

Recombinant Full Length Human ATXN1 Protein, fused to Flag-tag at C-terminus, was expressed in Mammalian cells.

Species

Human

Source

Mammalian Cells

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). Alternative splicing results in multiple transcript variants, with one variant encoding

 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

multiple distinct proteins, ATXN1 and Alt-ATXN1, due to the use of overlapping alternate reading frames.

Form 25 mM Tris HCl, pH 7.3, 100 mM glycine, 10% glycerol.

Molecular Mass 86.7 kDa

AA Sequence

MKSNQERSNECLPPKKREIPATSRSSSEKAPTLPSDNHRVEGTAWLPGNPGGRGH
GGGRHGPGAGTSVELG LQQGIGLHKALSTGLDYSPPSAPRSVPVATTLPAAYATPQP
GTPVSPVQYAHLPHTFQFIGSSQYSGTYA SFIPSQLIPPTANPVTSAVASAAGATTP
SQRSQLEAYSTLLANMGSLSQTPGHKAEQQQQQQQQQQQQHQ HQQQQQQQQQ
QQQQQHLSRAPGLITPGSPPPAQQNQYVHISSSPQNTGRTASPPAIPVHLHPHQMTI
PHT LTLGPPSQVVMQYADSGSHFVPREATKKAESSRLQQAIQAKEVLNGEMEKSR
RYGAPSSADLGLGKAGGK SVPHPYESRHVVVHPSPSDYSSRDPSGVRASVMVLP
NSNTPAADLEVQQATHREASPSTLNDKSGLHLGK PGHRSYALSPHTVIQTTHSASE
PLPVGLPATAFYAGTQPPVIGYLSGQQQAITIYAGSLPQHLVIPGTQPL LIPVGSTDME
ASGAAPAVTSSPQFAAVPHTFVTTALPKSENFNPEALVTQAAYPAMVQAQIHLPPVV
QSV ASPAAAPPTLPPYFMKGSIIQLANGELKKVEDLKTEDFIQSAEISNDLKIDSSTVE
RIEDSHSPGVAVIQ FAVGEHRAQVSVEVLVEYPPFFVFGQGWSGCCPERTSQLFDLP
CSKLSVGDVCISLTLKLNKNGSVKKGQP VDPASVLLKHSKADGLAGSRHRYAEQEN
GINQGSAQMLSENGELKFPEKMGLPAAPFLTKEPSKPAATR
KRRWSAPESRKLEKSEDEPPLTLPKPSLIPQEVKICIEGRSNVKGKTRTRPLEQKLISE
EDLAANDILDYKDDDDKV

Purity > 80% as determined by SDS-PAGE and Coomassie blue staining.

Stability Stable for 12 months from the date of receipt of the product under proper storage and handling conditions. Avoid repeated freeze-thaw cycles.

Storage Store at -80 centigrade.

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Concentration >50 ug/mL as determined by microplate BCA method.

Preparation Recombinant protein was captured through anti-DDK affinity column followed by conventional chromatography steps.

Full Length Full L.

GENE INFORMATION

Gene Name [ATXN1 ataxin 1 \[Homo sapiens \(human\) \]](#)

Official Symbol [ATXN1](#)

Synonyms ATX1; SCA1; D6S504E

Gene ID [6310](#)

mRNA Refseq [NM_000332.4](#)

Protein Refseq [NP_000323.2](#)

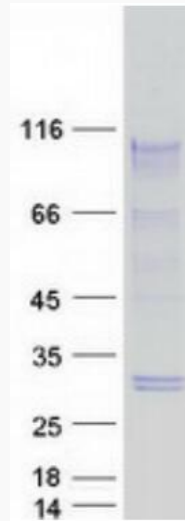
MIM [601556](#)

UniProt ID [P54253](#)

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Coomassie blue staining of purified ATXN1 protein.

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