

# Recombinant Human ATXN1 Protein, Myc/DDK-tagged, C13 and N15-labeled

**Cat. No.** ATXN1-1994H    **Lot. No.** (See product label)

## SPECIFICATION

### Product Overview

ATXN1 MS Standard C13 and N15-labeled recombinant protein (NP\_000323) with a C-terminal MYC/DDK tag, was expressed in HEK293 cells.

### Species

Human

### Source

HEK293

### 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

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

**Molecular Mass** 86.9 kDa

**AA Sequence**

MKSNQERSNECLPPKKREIPATSRSSSEKAPTLPDNRHVEGTAWLPGNPGGRGH  
 GGGRHGPAAGTSVELGLQQGIGLHKALSTGLDYSPPSAPRSVPVATTLPAAYATPQP  
 GTPVSPVQYAHLPHTFQFIGSSQYSGTYASFIPSQLIPPTANPVTSAVASAAGATTPS  
 QRSQLEAYSTLLANMGSLSQTPGHKAEQQQQQQQQQQQQHQHQHQHQHQHQHQHQ  
 QQQQHLSRAPGLITPGSPPPAQQNQYVHISSSPQNTGRTASPPAIPVHLHPHQTMIP  
 HTLTLGPPSQVVMQYADSGSHFVPREATKKAESSRLQQAIAKEVLNGEMEKSRRY  
 GAPSSADLGLGKAGGKSVPHPYESRHVVVHPSPSDYSSRDPSGVRASVMVLPNSN  
 TPAADLEVQQATHREASPSTLNDKSGHLHGKPGHRSYALSPHTVIQTTHSASEPLPV  
 GLPATAFYAGTQPPVIGYLSGQQQAITIYAGSLPQHLVIPGTQPLLIPVGSTDMEASGA  
 APAIVTSSPQFAAVPHTFVTTALPKSENFNPEALVTQAAYPAMVQAQIHLPVVQSVA  
 SPAAAPPTLPPYFMKGSIIQLANGELKKVEDLKTEDFIQSAEISNDLKIDSSTVERIEDS  
 HSPGVAVIQFAVGEHRAQVSVEVLVEYPFFVFGQGWSSCCPERTSQLFDLPCSKLS  
 VGDVCISLTLKLNKNGSVKKGQPVDPASVLLKHSKADGLAGSRHRYAEQENGINQG  
 SAQMLSENGELKFPEKMGLPAAPFLTKIEPSKPAATRKRWRSAPESTRKLEKSEDEP  
 PLTLPKPSLIPQEVKICIEGRSNVVGKTRTRPLEQKLISEEDLAANDILDYKDDDDKV

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

**Stability** Stable for 3 months from receipt of products under proper storage and handling conditions.

**Storage** Store at -80 centigrade. Avoid repeated freeze-thaw cycles.

**Concentration** 50 µg/mL as determined by BCA

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**Storage Buffer** 100 mM glycine, 25 mM Tris-HCl, pH 7.3.

## GENE INFORMATION

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

**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](#)

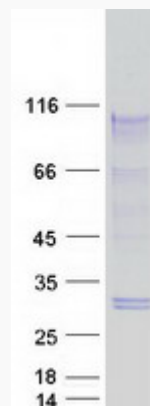
**mRNA Refseq** [NM\\_000332](#)

**Protein Refseq** [NP\\_000323](#)

**MIM** [601556](#)

**UniProt ID** [P54253](#)

## SDS-PAGE



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