

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

MKSNQERSNECLPPKKREIPATSRSSSEKAPTLPSDNHRVEGTAWLPGNPGGRGH
GGGRHGPAGTSVELGLQQGIGLHKALSTGLDYSPPSAPRSVPVATTLPAAYATPQP
GTPVSPVQYAHLPHTFQFIGSSQYSGTYASFIPSQLIPTANPVTSAVASAAGATTPS
QRSQLEAYSTLLANMGSLSQTPGHKAEQQQQQQQQQQQQHQQQQQQQQQQQ
QQQQHLSRAPGLITPGSPPPAQQNQYVHISSSPQNTGRTASPPAIPVHLHPHQTMI
HTLTGPPSQVVMQYADSGSHFVPREATKKAESSRLQQAIAKEVLNGEMEKSRRY
GAPSSADLGLGKAGGKSVPHYESRHHVVHPSPSDYSSRDPSGVRASVMVLPNSN
TPAADLEVQQATHREASPSTLNDKSGHLHGKPGHRSYALSPHTVIQTTHSASEPLPV
GLPATAFYAGTQPPVIGYLSGQQQAITYAGSLPQHLLVIPGTQPLLIPVGSTDMEASGA
APAITSSPQFAAVPHTFVTTALPKSENFNPEALVTQAAYPAMVQAQIHLPPVQSVA
SPAAAPPTLPPYFMKGSIIQLANGELKKVEDLKTEDFIQSAEISNDLKIDSSTVERIEDS
HSPGVAVIQFAVGEHRAQVSVEVLVEYPFFVFGQGWSSCCPERTSQLFDLPCKLS
VGDVCISLTLKLNKNGSVKKGQPVDPASVLLKHSKADGLAGSRHRYAEQENGINQG
SAQMLSENGELKFPEKMGLPAAPFLTKEPSKPAATRKRWSAPESRKLEKSEDEP
PLTLPKPSLIPQEVKICIEGRSNVGKTRTRPLEQKLISEEDLAANDILDYKDDDDKV

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](#)

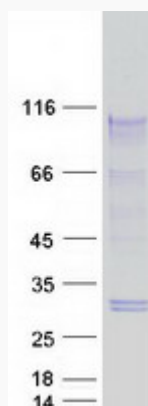
mRNA Refseq [NM_000332](#)

Protein Refseq [NP_000323](#)

MIM [601556](#)

UniProt ID [P54253](#)

SDS-PAGE



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