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	MKSNQERSNECLPPKKREIPATSRSSEEKAPTLPSDNHRVEGTAWLPGNPGGRGH GGGRHGPAGTSVELGLQQGIGLHKALSTGLDYSPPSAPRSVPVATTLPAAYATPQP GTPVSPVQYAHLPHTFQFIGSSQYSGTYASFIPSQLIPPTANPVTSAVASAAGATTPS QRSQLEAYSTLLANMGSLSQTPGHKAEQQQQQQQQQQQHQHQQQQQQQQQQ QQQQHLSRAPGLITPGSPPPAQQNQYVHISSSPQNTGRTASPPAIPVHLHPHQTMIP HTLTLGPPSQVVMQYADSGSHFVPREATKKAESSRLQQAIQAKEVLNGEMEKSRRY GAPSSADLGLGKAGGKSVPHPYESRHVVVHPSPSDYSSRDPSGVRASVMVLPNSN TPAADLEVQQATHREASPSTLNDKSGLHLGKPGHRSYALSPHTVIQTTHSASEPLPV GLPATAFYAGTQPPVIGYLSGQQQAITYAGSLPQHLVIPGTQPLLIPVGSTDMEASGA APAIVTSSPQFAAVPHTFVTTALPKSENFNPEALVTQAAYPAMVQAQIHLPVVQSVA SPAAAPPTLPPYFMKGSIIQLANGELKKVEDLKTEDFIQSAEISNDLKIDSSTVERIEDS HSPGVAVIQFAVGEHRAQVSVEVLVEYPFFVFGQGWSSCCPERTSQLFDLPCSKLS VGDVCISLTLKNLKNGSVKKGQPVDPASVLLKHSKADGLAGSRHRYAEQENGINQG SAQMLSENGELKFPEKMGLPAAPFLTKIEPSKPAATRKRRWSAPESRKLEKSEDEP 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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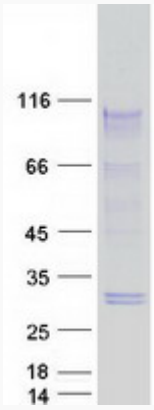
Storage Buffer	100 mM glycine, 25 mM Tris-HCl, pH 7.3.

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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