

## Product datasheet for PH326185

### Ataxin 1 (ATXN1) (NM\_001128164) Human Mass Spec Standard

#### Product data:

Product Type:	Mass Spec Standards
Description:	ATXN1 MS Standard C13 and N15-labeled recombinant protein (NP_001121636)
Species:	Human
Expression Host:	HEK293
Expression cDNA Clone or AA Sequence:	RC226185
Predicted MW:	86.9 kDa
Protein Sequence:	>RC226185 protein sequence Red=Cloning site Green=Tags(s)

MKSNQERSNECLPPKKREIPATSRSSSEKAPTLPDNRHVEGTAWLPGNPGGRGHGGRRHGHPAGTSVELG  
LQQGIGLHKALSTGLDYSPPSAPRSVPVATTLPAAYATPQPGTPVSPVQYAHLPHTFQFIGSSQYSGTYA  
SFIPSQLIPPTANPVTSAVASAAGATTPSQRSQLEAYSTLLANMGSLSQTPGHKAEQQQQQQQQQQHQ  
HQQQQQQQQQQQQHL SRAPGLITPGSPPPAQQNQYVHISSSPQNTGRTASPPAIPVHLHPHQTMIPIHT  
LTLGPPSQVVMQYADSGSHFVPREATKKAESSRLQQAIAKEVLNGEMEKSRRYGAPSSADLGLGKAGGK  
SVPHPYESRHVVVHPSPSDYSSRDPSGVRASVMVLPNSNTPAADLEVQQATHREASPSTLNDKSGLHLGK  
PGHRSYALSPHTVIQTTHSASEPLVGLPATAFYAGTQPPVIGYLSGQQQAITYAGSLPQHLVIPGTQPL  
LIPVGSTDMEASGAAPAIVTSSPQFAAVPHTFVTTALPKSENFNPEALVTQAAYPAMVQAQIHLVAVQSV  
ASPAAPPTLPPYFMKGSIIQLANGELKKVEDLKTEDFIQSAEISNDLKIDSSTVERIEDSHSPGVAVIQ  
FAVGEHRAQVSVEVLVEYPFFVFGQGWSSCCPERTSQLFDLPCSKLSVGDVCISLTLKNLKNGSVKKGQP  
VDPASVLLKHSKADGLAGSRHRYAEQENINQGSQMLSENGELKFPEKMGLPAAPFLTKIEPSKPAATR  
KRRWSAPESRKLEKSEDEPPLTLPKPSLIPQEVKICIEGRSNVGK

TRTRPLEQKLI SEEDLAANDILDYKDDDDKV

Tag:	C-Myc/DDK
Purity:	> 80% as determined by SDS-PAGE and Coomassie blue staining
Concentration:	>0.05 µg/µL as determined by microplate BCA method
Labeling Method:	Labeled with [U- <sup>13</sup> C <sub>6</sub> , <sup>15</sup> N <sub>4</sub> ]-L-Arginine and [U- <sup>13</sup> C <sub>6</sub> , <sup>15</sup> N <sub>2</sub> ]-L-Lysine
Buffer:	25 mM Tris-HCl, 100 mM glycine, pH 7.3
Storage:	Store at -80°C. Avoid repeated freeze-thaw cycles.
Stability:	Stable for 3 months from receipt of products under proper storage and handling conditions.
RefSeq:	<a href="#">NP_001121636</a>

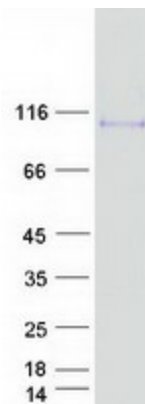


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RefSeq Size:	10587
RefSeq ORF:	2445
Synonyms:	ATX1; D6S504E; SCA1
Locus ID:	6310
UniProt ID:	<a href="#">P54253</a> , <a href="#">Q96FF1</a>
Cytogenetics:	6p22.3

**Summary:** 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 multiple distinct proteins, ATXN1 and Alt-ATXN1, due to the use of overlapping alternate reading frames. [provided by RefSeq, Nov 2017]

## Product images:



Coomassie blue staining of purified ATXN1 protein (Cat# [TP326185]). The protein was produced from HEK293T cells transfected with ATXN1 cDNA clone (Cat# [RC226185]) using MegaTran 2.0 (Cat# [TT210002]).