

Product datasheet for PH322862

OriGene Technologies, Inc.

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Ataxin 1 (ATXN1) (NM 000332) Human Mass Spec Standard

Product data:

Product Type: Mass Spec Standards

ATXN1 MS Standard C13 and N15-labeled recombinant protein (NP_000323) **Description:**

Species: Human **HEK293 Expression Host: Expression cDNA Clone**

or AA Sequence:

RC222862

Predicted MW: 86.9 kDa

>RC222862 protein sequence **Protein Sequence:**

Red=Cloning site Green=Tags(s)

MKSNOERSNECLPPKKREIPATSRSSEEKAPTLPSDNHRVEGTAWLPGNPGGRGHGGGRHGPAGTSVELG LQQGIGLHKALSTGLDYSPPSAPRSVPVATTLPAAYATPQPGTPVSPVQYAHLPHTFQFIGSSQYSGTYA SFIPSQLIPPTANPVTSAVASAAGATTPSQRSQLEAYSTLLANMGSLSQTPGHKAEQQQQQQQQQQQQQQHQ HQQQQQQQQQQQQQHLSRAPGLITPGSPPPAQQNQYVHISSSPQNTGRTASPPAIPVHLHPHQTMIPHT LTLGPPSQVVMQYADSGSHFVPREATKKAESSRLQQAIQAKEVLNGEMEKSRRYGAPSSADLGLGKAGGK SVPHPYESRHVVVHPSPSDYSSRDPSGVRASVMVLPNSNTPAADLEVQQATHREASPSTLNDKSGLHLGK PGHRSYALSPHTVIQTTHSASEPLPVGLPATAFYAGTQPPVIGYLSGQQQAITYAGSLPQHLVIPGTQPL LIPVGSTDMEASGAAPAIVTSSPQFAAVPHTFVTTALPKSENFNPEALVTQAAYPAMVQAQIHLPVVQSV ASPAAAPPTLPPYFMKGSIIQLANGELKKVEDLKTEDFIQSAEISNDLKIDSSTVERIEDSHSPGVAVIQ FAVGEHRAQVSVEVLVEYPFFVFGQGWSSCCPERTSQLFDLPCSKLSVGDVCISLTLKNLKNGSVKKGQP VDPASVLLKHSKADGLAGSRHRYAEQENGINQGSAQMLSENGELKFPEKMGLPAAPFLTKIEPSKPAATR

KRRWSAPESRKLEKSEDEPPLTLPKPSLIPQEVKICIEGRSNVGK

TRTRPLEOKLISEEDLAANDILDYKDDDDKV

Tag: C-Myc/DDK

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

>0.05 µg/µL as determined by microplate BCA method **Concentration:**

Labeling Method: Labeled with [U- 13C6, 15N4]-L-Arginine and [U- 13C6, 15N2]-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: NP 000323





RefSeq Size: 10636

RefSeq ORF: 2445

Synonyms: ATX1; D6S504E; SCA1

Locus ID: 6310

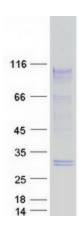
UniProt ID: <u>P54253</u>, <u>Q96FF1</u>

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# [TP322862]). The protein was produced from HEK293T cells transfected with ATXN1 cDNA clone (Cat# [RC222862]) using MegaTran 2.0 (Cat# [TT210002]).