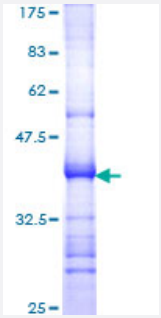
# ATXN1 (Human) Recombinant Protein (Q01)

Catalog # H00006310-Q01 Size 25 ug, 10 ug

## Applications




## Specification

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|---|---|
| **Product Description** | Human ATXN1 partial ORF ( NP_000323, 576 a.a. - 675 a.a.) recombinant protein with GST-tag at N-terminal. |
| **Sequence** | KGSIIQLANGELKKVEDLKTEDFIQSAEISNDLKIDSSTVERIEDSHSPGVAVIQFAVGEHRAQVSVE VLVEYPFFVFGQGWSSCCPERTSQLFDLPCSK |
| **Host** | Wheat Germ (in vitro) |
| **Theoretical MW (kDa)** | 36.74 |
| **Interspecies Antigen Sequence** | Mouse (92); Rat (100) |
| **Preparation Method** | *in vitro* wheat germ expression system |
| **Purification** | Glutathione Sepharose 4 Fast Flow |
| **Quality Control Testing** | 12.5% SDS-PAGE Stained with Coomassie Blue. |
| **Storage Buffer** | 50 mM Tris-HCl, 10 mM reduced Glutathione, pH=8.0 in the elution buffer. |
| **Storage Instruction** | Store at -80°C. Aliquot to avoid repeated freezing and thawing. |
| **Note** | Best use within three months from the date of receipt of this protein. |

## Applications

- Enzyme-linked Immunoabsorbent Assay
- Western Blot (Recombinant protein)
- Antibody Production
- Protein Array

## Gene Info — ATXN1

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|---|---|
| **Entrez GeneID** | 6310 |
| **GeneBank Accession#** | NM_000332 |
| **Protein Accession#** | NP_000323 |
| **Gene Name** | ATXN1 |
| **Gene Alias** | ATX1, D6S504E, SCA1 |
| **Gene Description** | ataxin 1 |
| **Omim ID** | 164400 601556 |
| **Gene Ontology** | Hyperlink |
| **Gene Summary** | The autosomal dominant cerebellar ataxias (ADCA) are a heterogeneous group of neurodegener ative disorders characterized by progressive degeneration of the cerebellum, brain stem and spin al cord. Clinically, ADCA has been divided into three groups: ADCA types I-III. ADCAI is genetical ly 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 degen eration (SCA7), and ADCAIII often referred to as the `pure' cerebellar syndrome (SCA5), are mos t likely homogeneous disorders. Several SCA genes have been cloned and shown to contain CA G repeats in their coding regions. ADCA is caused by the expansion of the CAG repeats, produci ng an elongated polyglutamine tract in the corresponding protein. The expanded repeats are varia ble in size and unstable, usually increasing in size when transmitted to successive generations. Th e function of the ataxins is not known. This locus has been mapped to chromosome 6, and it has b een determined that the diseased allele contains 41-81 CAG repeats, compared to 6-39 in the no rmal allele. At least two transcript variants encoding the same protein have been found for this gen e. [provided by RefSeq |
| **Other Designations** | OTTHUMP00000016065\|OTTHUMP00000039306\|olivopontocerebellar ataxia 1, autosomal dom inant\|spinocerebellar ataxia 1 (olivopontocerebellar ataxia 1, autosomal dominant, ataxin 1) |

## Publication Reference

- Scaffold function of long non-coding RNA HOTAIR in protein ubiquitination.

  Yoon JH, Abdelmohsen K, Kim J, Yang X, Martindale JL, Tominaga-Yamanaka K, White EJ, Orjalo AV, Rinn JL, Kreft SG, Wilson GM, Gorospe M.

  Nature Communications 2013 Dec; 4:2939.

  Application: Ubiquitination assay, Human, HeLa cells

## Disease

- Alzheimer Disease
- Bipolar Disorder
- Cerebellar Ataxia
- Chronic Disease
- Cognition
- Dementia
- Diseases in Twins
- Epilepsy
- Fragile X syndrome
- Friedreich Ataxia
- Genetic Predisposition to Disease
- Genomic Instability
- Huntington disease
- Machado-Joseph Disease
- Muscular Atrophy
- Myoclonic Epilepsies
- Myotonic dystrophy
- Neurodegenerative Diseases

- Parkinson disease
- Restless Legs Syndrome
- Schizophrenia
- Spinal muscular atrophy
- Spinocerebellar ataxia
- Spinocerebellar Ataxias
- Tobacco Use Disorder