

Recombinant Human ATXN2, His-tagged

Cat. No. ATXN2-10068H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant Human ATXN2 protein, fused to His-tag, was expressed in E.coli and purified by Ni-sepharose.
Species	Human
Source	E.coli
ProteinLength	540-889a.a.
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. Defects in this gene are the cause of spinocerebellar ataxia type 2 (SCA2). SCA2 belongs to the autosomal dominant cerebellar ataxias type I (ADCA I) which are characterized by cerebellar ataxia in combination with additional clinical features like optic atrophy, ophthalmoplegia, bulbar and extrapyramidal signs, peripheral neuropathy and dementia. SCA2 is caused by expansion of a CAG repeat in the coding region of this gene. This locus has been mapped to chromosome 12, and it has been determined that the diseased allele contains 37-50 CAG repeats, compared to 17-29 in the normal allele. Longer expansions result in earlier onset of the disease. Alternatively spliced transcript variants encoding different isoforms have been identified but their full length sequence has not been determined.
Storage	The protein is stored in PBS buffer at -20°C. Avoid repeated freezing and thawing

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cycles.

Storage Buffer

1M PBS (58mM Na₂HPO₄, 17mM NaH₂PO₄, 68mM NaCl, pH8.) added with 300mM Imidazole and 0.7% Sarcosyl, 15% glycerol.

GENE INFORMATION

Gene Name

[ATXN2 ataxin 2 \[Homo sapiens \]](#)

Official Symbol

ATXN2

Synonyms

ATXN2; ataxin 2; SCA2, spinocerebellar ataxia 2 (olivopontocerebellar ataxia 2, autosomal dominant, ataxin 2) , TNRC13; ataxin-2; ATX2; trinucleotide repeat containing 13; spinocerebellar ataxia type 2 protein; trinucleotide repeat-containing gene 13 protein; SCA2; TNRC13; FLJ46772;

Gene ID

[6311](#)

mRNA Refseq

[NM_002973](#)

Protein Refseq

[NP_002964](#)

MIM

[601517](#)

UniProt ID

[Q99700](#)

**Chromosome
Location**

12q23-q24.1

Function

RNA binding; protein C-terminus binding; protein binding;

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