

ATXN1

Reactivity: Human Mouse Rat

Tested applications: WB IHC IF

Recommended Dilution: WB 1:500 - 1:2000 IHC 1:50 - 1:200 IF 1:10 - 1:100

Calculated MW: 87kDa

Observed MW: Refer to Figures

Immunogen:

Recombinant protein of human ATXN1

Storage Buffer:

Store at -20. Avoid freeze / thaw cycles. Buffer: PBS with 0.02% sodium azide, 50% glycerol, pH7.3.

Synonym:

ATX1; SCA1; D6S504E

Catalog #: A6217

Antibody Type:

Polyclonal Antibody

Species: Rabbit

Gene ID: 6310

Isotype: IgG

Swiss Prot: P54253

Purity: Affinity purification

For research use only.

Background:

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 41-81 CAG repeats, compared to 6-39 in the normal allele, and is associated with spinocerebellar ataxia type 1 (SCA1). At least two transcript variants encoding the same protein have been found for this gene.

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