



Human ATXN7 peptide (DAG-P0190)

This product is for research use only and is not intended for diagnostic use.

PRODUCT INFORMATION

Antigen 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. 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. This locus has been mapped to chromosome 3, and it has been determined that the diseased allele associated with spinocerebellar ataxia-7 contains 38-130 CAG repeats (near the N-terminus), compared to 7-17 in the normal allele. The encoded protein is a component of the SPT3/TAF9/GCN5 acetyltransferase (STAGA) and TBP-free TAF-containing (TFTC) chromatin remodeling complexes, and it thus plays a role in transcriptional regulation. Alternative splicing results in multiple transcript variants. [provided by RefSeq, Apr 2010]

Purity	> 90 % by SDS-PAGE.>70% pure.
Conjugate	Unconjugated
Applications	Neut, WB
Format	Liquid
Buffer	Double distilled water
Preservative	None
Storage	Shipped at 4°C. Upon delivery aliquot and store at -20°C. Avoid freeze / thaw cycles. Double distilled water

GENE INFORMATION

Gene Name	ATXN7 ataxin 7 [Homo sapiens (human)]
Official Symbol	ATXN7
Synonyms	ATXN7; ataxin 7; SCA7; OPCA3; ADCAll; ataxin-7; spinocerebellar ataxia type 7 protein;
Entrez Gene ID	6314
mRNA Refseq	NM_000333.3
Protein Refseq	NP_000324.1
UniProt ID	O15265
Chromosome Location	3p21.1-p12
Pathway	Chromatin modifying enzymes, organism-specific biosystem; Chromatin organization, organism-specific biosystem; HATs acetylate histones, organism-specific biosystem;
Function	chromatin binding; protein binding;