

Product datasheet for **KN222862LP**

Ataxin 1 (ATXN1) Human Gene Knockout Kit (CRISPR)

Product data:

Product Type:	Knockout Kits (CRISPR)
Format:	2 gRNA vectors, 1 Luciferase-Puro donor, 1 scramble control
Donor DNA:	Luciferase-Puro
Symbol:	Ataxin 1
Locus ID:	6310
Components:	KN222862G1 , Ataxin 1 gRNA vector 1 in pCas-Guide CRISPR vector (GE100002) KN222862G2 , Ataxin 1 gRNA vector 2 in pCas-Guide CRISPR vector (GE100002) KN222862LPD , donor DNA containing left and right homologous arms and Luciferase-Puro functional cassette. GE100003 , scramble sequence in pCas-Guide vector
Disclaimer:	These products are manufactured and supplied by OriGene under license from ERS. The kit is designed based on the best knowledge of CRISPR technology. The system has been functionally validated for knocking-in the cassette downstream the native promoter. The efficiency of the knock-out varies due to the nature of the biology and the complexity of the experimental process.
RefSeq:	NM_000332 , NM_001128164 , NM_001357857 , NR_152111 , NR_152112 , NR_152113 , NR_152114 , N52856
UniProt ID:	P54253
Synonyms:	ATX1; D6S504E; SCA1



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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. ADCA I is genetically heterogeneous, with five genetic loci, designated spinocerebellar ataxia (SCA) 1, 2, 3, 4 and 6, being assigned to five different chromosomes. ADCA II, which always presents with retinal degeneration (SCA7), and ADCA III 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:
