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Product datasheet for RC222341L1V

ATX2 (ATXN2) (NM_002973) Human Tagged ORF Clone Lentiviral Particle

Product data:

Product Type:	Lentiviral Particles
Product Name:	ATX2 (ATXN2) (NM_002973) Human Tagged ORF Clone Lentiviral Particle
Symbol:	ATX2
Synonyms:	ATX2; SCA2; TNRC13
Mammalian Cell Selection:	None
Vector:	pLenti-C-Myc-DDK (PS100064)
Tag:	Myc-DDK
ACCN:	NM_002973
ORF Size:	3939 bp
ORF Nucleotide Sequence:	The ORF insert of this clone is exactly the same as(RC222341).
OTI Disclaimer:	The molecular sequence of this clone aligns with the gene accession number as a point of reference only. However, individual transcript sequences of the same gene can differ through naturally occurring variations (e.g. polymorphisms), each with its own valid existence. This clone is substantially in agreement with the reference, but a complete review of all prevailing variants is recommended prior to use. More info
OTI Annotation:	This clone was engineered to express the complete ORF with an expression tag. Expression varies depending on the nature of the gene.
RefSeq:	NM_002973.3, NP_002964.3
RefSeq Size:	4702 bp
RefSeq ORF:	3462 bp
Locus ID:	6311
UniProt ID:	Q99700
Cytogenetics:	12q24.12
MW:	140.1 kDa

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Gene Summary: This gene belongs to a group of genes that is associated with microsatellite-expansion diseases, a class of neurological and neuromuscular disorders caused by expansion of short stretches of repetitive DNA. The protein encoded by this gene has two globular domains near the N-terminus, one of which contains a clathrin-mediated trans-Golgi signal and an endoplasmic reticulum exit signal. The encoded cytoplasmic protein localizes to the endoplasmic reticulum and plasma membrane, is involved in endocytosis, and modulates mTOR signals, modifying ribosomal translation and mitochondrial function. The N-terminal region of the protein contains a polyglutamine tract of 14-31 residues that can be expanded in the pathogenic state to 32-200 residues. Intermediate length expansions of this tract increase susceptibility to amyotrophic lateral sclerosis, while long expansions of this tract result in spinocerebellar ataxia-2, an autosomal-dominantly inherited, neurodegenerative disorder. Genome-wide association studies indicate that loss-of-function mutations in this gene may be associated with susceptibility to type I diabetes, obesity and hypertension. Alternative splicing results in multiple transcript variants. [provided by RefSeq, Nov 2016]