

## CAB16666

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**Product Information**

<b>Product SKU:</b>	CAB16666	<b>Gene ID:</b>	6311	<b>Size:</b>	20uL, 100uL
<b>Clone No:</b>	-	<b>Host Species:</b>	Rabbit	<b>Reactivity:</b>	Human,Mouse,Rat

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**Additional Information**

<b>Observed MW:</b>	150kDa	<b>Conjugate:</b>	Unconjugated
<b>Calculated MW:</b>	140kDa	<b>Isotype:</b>	IgG

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**Immunogen Information**

**Background:** 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.

**Recommended Dilution:** WB,1:500 - 1:2000 IHC-P,1:50 - 1:200 IF/ICC,1:50 - 1:200

**Synonyms:** ATX2; SCA2; TNRC13; ATXN2

**Purification Method:** Affinity purification

**Immunogen:** A synthetic peptide corresponding to a sequence within amino acids 1250-1331 of human ATXN2 (NP\_002964.3).

**Storage:** Store at -20°C. Avoid freeze / thaw cycles.Buffer: PBS with 0.01% thimerosal,50% glycerol,pH7.3.