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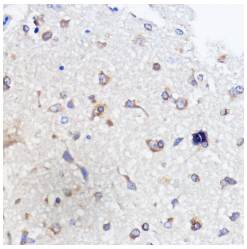
ATXN2 Polyclonal Antibody

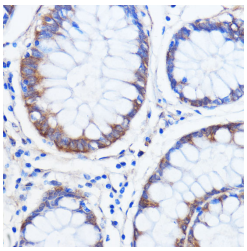
Catalog No.	E-AB-66891	**Reactivity**	H,M,R
Storage	Store at -20°C. Avoid freeze / thaw cycles.	**Host**	Rabbit
Applications	IHC,IF	**Isotype**	IgG

Note: Centrifuge before opening to ensure complete recovery of vial contents.

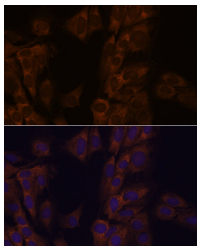
Images



Immunohistochemistry of paraffin-embedded Rat brain using ATXN2 Polyclonal Antibody at dilution of 1:100 (40x lens).



Immunohistochemistry of paraffin-embedded Human colon carcinoma using ATXN2 Polyclonal Antibody at dilution of 1:100 (40x lens).



Immunofluorescence analysis of C6 cells using ATXN2 Polyclonal Antibody at dilution of 1:100. Blue: DAPI for nuclear staining.

Immunogen Information

Immunogen	A synthetic peptide of human ATXN2
GeneID	6311
Swissprot	Q99700
Synonyms	ATXN2,ATX2,SCA2,TNRC13,ataxin-2

Product Information

Buffer	PBS with 0.02% sodium azide, 50% glycerol, pH7.3.
Purify	Affinity purification
Dilution	IHC 1:50-1:200 IF 1:50-1:200

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.