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AFG3L2 Rabbit pAb

Catalog # AP99761

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	Q9Y4W6
Other Accession	Q9Y4W6
Reactivity	Rat, Mouse
Predicted	Pig, Human, Chicken, Dog, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	88584
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human AFG3L2
Epitope Specificity	531-600/797
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SIMILARITY	In the N-terminal section; belongs to the AAA ATPase family. In the C-terminal section; belongs to the peptidase M41 family.
SUBUNIT	Homooligomer. Interacts with SPG7; the interaction is required for the efficient assembly of mitochondrial complex I.
DISEASE	Defects in AFG3L2 are the cause of spinocerebellar ataxia type 28 (SCA28) [MIM:610246]. It is a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with variable involvement of the brainstem and spinal cord. SCA28 is an autosomal dominant cerebellar ataxia (ADCA) with a slow progressive course and no evidence of sensory involvement or cognitive impairment. Defects in AFG3L2 are the cause of spastic ataxia autosomal recessive type 5 (SPAX5) [MIM:614487]. A neurodegenerative disorder characterized by early onset spasticity, peripheral neuropathy, ptosis, oculomotor apraxia, dystonia, cerebellar atrophy, and progressive myoclonic epilepsy.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	AFG3L2 is a multi-pass membrane metalloprotease that contains one AAA (ATPase associated with diverse cellular activities) domain, a zinc-dependent binding motif, an RNA-binding region and an ATP/GTP binding site. Localizing to the mitochondrial membrane, AFG3L2 is ubiquitously expressed with highest expression levels in skeletal muscle and heart. AFG3L2 shares 69% similarity with the yeast Afg3 protein and 49% similarity with Paraplegin, a protein of mitochondria that is thought to be involved in signal transduction and chaperone-like activities. In mitochondria, AFG3L2 forms a complex with Paraplegin that is believed to regulate essential protein quality control. Mutations in the gene encoding either one of these proteins can result in hereditary spastic paraplegia, a degenerative spinal cord disorder that is characterized by muscle spasms, stiffness in the legs and, in some cases, incontinence.

Additional Information

Gene ID	10939
Other Names	OPA12; SCA28; SPAX5; AFG32_HUMAN; AFG3L2; AFG3-like protein 2; Paraplegin-like protein; 3.4.24.-; 3.6.-.-; AFG3 like matrix AAA peptidase subunit 2; AFG3 (ATPase family gene 3, yeast)-like 2; spinocerebellar ataxia 28; AFG3 ATPase family member 3-like 2 (S. cerevisiae); AFG3 like AAA ATPase 2
Dilution	IHC-P=1:100-500,IHC-F=1:100-500,IF=1:100-500
Storage	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.

Protein Information

Name	AFG3L2 {ECO:0000303|PubMed:10395799, ECO:0000312|HGNC:HGNC:315}
Function	Catalytic component of the m-AAA protease, a protease that plays a key role in proteostasis of inner mitochondrial membrane proteins, and which is essential for axonal and neuron development (PubMed:19748354, PubMed:28396416, PubMed:29932645, PubMed:30683687, PubMed:31327635, PubMed:37917749, PubMed:38157846, PubMed:41075794). AFG3L2 possesses both ATPase and protease activities: the ATPase activity is required to unfold substrates, threading them into the internal proteolytic cavity for hydrolysis into small peptide fragments (PubMed:19748354, PubMed:31327635). The m-AAA protease carries out quality control in the inner membrane of the mitochondria by mediating degradation of mistranslated or misfolded polypeptides (PubMed:26504172, PubMed:30683687, PubMed:34718584). The m-AAA protease complex also promotes the processing and maturation of mitochondrial proteins, such as MRPL32/bL32m, PINK1 and SP7 (PubMed:22354088, PubMed:29932645, PubMed:30252181). Mediates protein maturation of the mitochondrial ribosomal subunit MRPL32/bL32m by catalyzing the cleavage of the presequence of MRPL32/bL32m prior to assembly into the mitochondrial ribosome (PubMed:29932645). Required for SPG7 maturation into its active mature form after SPG7 cleavage by mitochondrial- processing peptidase (MPP) (PubMed:30252181). Required for the maturation of PINK1 into its 52kDa mature form after its cleavage by mitochondrial-processing peptidase (MPP) (PubMed:22354088). Acts as a regulator of calcium in neurons by mediating degradation of SMDT1/EMRE before its assembly with the uniporter complex, limiting the availability of SMDT1/EMRE for MCU assembly and promoting efficient assembly of gatekeeper subunits with MCU (PubMed:27642048, PubMed:28396416). Promotes the proteolytic degradation of GHITM upon hyperpolarization of mitochondria: progressive GHITM degradation leads to respiratory complex I degradation and broad reshaping of the mitochondrial proteome by AFG3L2 (PubMed:35912435). Also acts as a regulator of mitochondrial glutathione homeostasis by mediating cleavage and degradation of SLC25A39 (PubMed:37917749, PubMed:38157846). SLC25A39 cleavage is prevented when SLC25A39 binds iron-sulfur (PubMed:37917749, PubMed:38157846). Also acts as a regulator of carnitine biosynthesis by mediating cleavage and degradation of SLC25A45 (PubMed:41075794). Involved in the regulation of OMA1-dependent processing of OPA1 (PubMed:17615298, PubMed:29545505, PubMed:30252181, PubMed:30683687, PubMed:32600459). May act by

	mediating processing of OMA1 precursor, participating in OMA1 maturation (PubMed:29545505).
Cellular Location	Mitochondrion inner membrane; Multi-pass membrane protein
Tissue Location	Ubiquitous. Highly expressed in the cerebellar Purkinje cells.

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