

Rabbit Anti-FIG4/AP Conjugated antibody

SL11690R-AP

Product Name	Anti-FIG4/AP
Chinese Name	碱性磷酸酶（AP）标记的肌萎缩侧索硬化症相关蛋白 FIG4 抗体
Alias	5-bisphosphate 5-phosphatase; Fig4; FIG4_HUMAN; KIAA0274; ALS11; Phosphatidylinositol 3; hSac3; Phosphatidylinositol 3,5 bisphosphate 5 phosphatase; Polyphosphoinositide phosphatase; SAC domain containing protein 3; SAC domain-containing protein 3; SAC3.
Research Area	Cell biology Neurobiology Signal transduction
Immunogen Species	Rabbit
Clonality	Polyclonal
React Species	Rat(predicted:Human,Mouse,Chicken,Dog,Pig,Sheep,Monkey) IHC-P=1:100-500,IHC-F=1:100-500
Applications	not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight	104kDa
Form	Lyophilized or Liquid
Concentration	1mg/ml
immunogen	KLH conjugated synthetic peptide derived from human FIG4 (1-100aa)
Lsotype	IgG
Purification	affinity purified by Protein A
Storage Buffer	1M TBS(pH7.4) with 1% BSA, 3% Proclin300 and 50% Glycerol. Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20°C. When reconstituted in sterile pH 7.4 1M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.
Storage	
Product Detail	background: The protein encoded by this gene belongs to the SAC domain-containing protein gene family. The SAC domain, approximately 400 amino acids in length and consisting of seven conserved motifs, has been shown to possess phosphoinositide phosphatase activity. The yeast homolog, Sac1p, is involved in the regulation of various phosphoinositides, and affects diverse cellular

functions such as actin cytoskeleton organization, Golgi function, and maintenance of vacuole morphology. Membrane-bound phosphoinositides function as signaling molecules and play a key role in vesicle trafficking in eukaryotic cells. Mutations in this gene have been associated with Charcot-Marie-Tooth disease, type 4J. [provided by RefSeq, Jul 2008]

Function:

The PI(3,5)P2 regulatory complex regulates both the synthesis and turnover of phosphatidylinositol-3,5-bisphosphate (PtdIns(3,5)P2). In vitro, hydrolyzes all three D5-phosphorylated polyphosphoinositide substrates in the order PtdIns(4,5)P2 > PtdIns(3,5)P2 > PtdIns(3,4,5)P3. Plays a role in the biogenesis of endosome carrier vesicles (ECV) / multivesicular bodies (MVB) transport intermediates from early endosomes.

Subcellular Location:

Endosome membrane. Localization requires VAC14 and PIKFYVE.

DISEASE:

Defects in FIG4 are the cause of Charcot-Marie-Tooth disease type 4J (CMT4J) [MIM:611228]. CMT4J is a recessive demyelinating, severe form of Charcot-Marie-Tooth disease, the most common inherited disorder of the peripheral nervous system. Charcot-Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies characterized by severely reduced motor nerve conduction velocities (NCVs) (less than 38m/s) and segmental demyelination and remyelination, and primary peripheral axonal neuropathies characterized by normal or mildly reduced NCVs and chronic axonal degeneration and regeneration on nerve biopsy.

Defects in FIG4 are the cause of amyotrophic lateral sclerosis type 11 (ALS11) [MIM:612577]. ALS is a neurodegenerative disorder affecting upper motor neurons in the brain and lower motor neurons in the brain stem and spinal cord, resulting in fatal paralysis. Sensory abnormalities are absent. Death usually occurs within 2 to 5 years. The etiology of amyotrophic lateral sclerosis is likely to be multifactorial, involving both genetic and environmental factors. The disease is inherited in 5-10%.

Similarity:

Contains 1 SAC domain.

Database links:

[Entrez Gene: 9896](#) Human



[Entrez Gene: 103199](#) Mouse

[Entrez Gene: 309855](#) Rat

[Omim: 609390](#) Human

[SwissProt: Q92562](#) Human

[SwissProt: Q91WF7](#) Mouse

[Unigene: 529959](#) Human

[Unigene: 277242](#) Mouse

Important Note:

This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.