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Rabbit Anti-FIG4/FITC Conjugated antibody

SL11690R-FITC

Product Name:	Anti-FIG4/FITC
Chinese Name:	FITC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.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Chicken,Dog,Pig,Sheep,Monkey,
Applications:	ICC=1:50-200IF=1:50-200 not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	104kDa
Cellular localization:	The cell membrane
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:	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Storage:	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 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.
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.