10320 Camino Santa Fe, Suite G
San Diego, CA 92121
Tel: 858.875.1900 Fax: 858.875.1999

SH3TC2 Rabbit pAb

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Catalog # AP54429

Product Information

Application	WB, IHC-P, IHC-F, IF
Primary Accession	Q8TF17
Predicted	Rat, Pig, Human, Mouse, Rabbit, Horse
Host	Rabbit
Clonality	Polyclonal
Calculated MW	144777
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human SH3TC2
Epitope Specificity	851-950/1288
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
DISEASE	Defects in SH3TC2 are the cause of Charcot-Marie-Tooth disease type 4C (CMT4C) [MIM:601596]. CMT4C is a recessive 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 neuropathy and primary peripheral axonal neuropathy. Demyelinating CMT neuropathies are characterized by severely reduced nerve conduction velocities (less than 38 m/sec), segmental demyelination and remyelination with onion bulb formations on nerve biopsy, slowly progressive distal muscle atrophy and weakness, absent deep tendon reflexes, and hollow feet. By convention, autosomal recessive forms of demyelinating Charcot-Marie-Tooth disease are designated CMT4. CMT4C is characterized by onset in childhood, early-onset scoliosis and a distinct Schwann cell pathology.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	SH3TC2 (SH3 domain and tetratricopeptide repeats 2) is a 1,288 amino acid protein that contains one SH3 domain and eight TPR repeats. The SH3TC2 gene encodes a protein expressed in Schwann cells of peripheral nerves, and localized to the plasma membrane and to the perinuclear endocytic recycling compartment, suggesting a possible function in myelination and/or in regions of axoglial interactions. The SH3TC2 protein is expressed in adult heart, testis, spinal cord, and brain as well as in fetal brain and liver. Mild mononeuropathy of the median nerve (MNMN) is caused by heterozygous mutation in the SH3TC2 gene. Also, Charcot-Marie-Tooth disease type 4C (CMT4C) is a more severe neuropathy caused by homozygous or compound heterozygous mutation in the SH3TC2 gene. Existing as four alternatively spliced isoforms and containing 18 exons, the SH3TC2 gene is conserved in chimpanzee, dog, cow, mouse, rat, chicken and zebrafish, and maps to human chromosome 5q32.

Additional Information

Gene ID	79628
Other Names	SH3 domain and tetratricopeptide repeat-containing protein 2, SH3TC2 (HGNC:29427)
Dilution	WB=1:500-2000,IHC-P=1:100-500,IHC-F=1:100-500,ICC/IF=1:100-500,IF=1:100-500,ELISA=1:5000-10000
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	SH3TC2 (HGNC:29427)
Function	Is involved in nerve myelination and is required for the integrity of nodes of Ranvier (By similarity). It probably functions as a Rab effector in the regulation of endocytic recycling (PubMed:20028792, PubMed:20826437).
Cellular Location	Cell membrane {ECO:0000250|UniProtKB:Q80VA5}. Recycling endosome
Tissue Location	Strongly expressed in brain and spinal cord. Expressed at equal level in spinal cord and sciatic nerve. Weakly expressed in striated muscle.

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