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SCN4A Rabbit pAb

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Catalog # AP56648

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	P35499
Predicted	Rat, Human, Mouse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	208061
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human SCN4A
Epitope Specificity	301-400/1836
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SUBCELLULAR LOCATION	Membrane.
DISEASE	Defects in SCN4A are the cause of paramyotonia congenita of von Eulenburg (PMC) [MIM:168300]. PMC is an autosomal dominant channelopathy characterized by myotonia, increased by exposure to cold, intermittent flaccid paresis, not necessarily dependent on cold or myotonia, lability of serum potassium, nonprogressive nature and lack of atrophy or hypertrophy of muscles. In some patients, myotonia is not increased by cold exposure (paramyotonia without cold paralysis). Patients may have a combination phenotype of PMC and HYPP. Defects in SCN4A are a cause of periodic paralysis hypokalemic type 2 (HOKPP2) [MIM:613345]. It is an autosomal dominant disorder manifested by episodic flaccid generalized muscle weakness associated with falls of serum potassium levels. Defects in SCN4A are the cause of periodic paralysis hyperkalemic (HYPP) [MIM:170500]. HYPP is an autosomal dominant channelopathy characterized by episodic flaccid generalized muscle weakness associated with high levels of serum potassium. Concurrence of myotonia is found in HYPP patients. Defects in SCN4A are the cause of periodic paralysis normokalemic (NKPP) [MIM:170500]. NKPP is a disorder closely related to hyperkalemic periodic paralysis, but marked by a lack of alterations in potassium levels during attacks of muscle weakness. Defects in SCN4A are the cause of myotonia SCN4A-related (MYOSCN4A) [MIM:608390]. Myotonia is characterized by sustained muscle tensing that prevents muscles from relaxing normally. Myotonia causes muscle stiffness that can interfere with movement. In some people the stiffness is very mild, while in other cases it may be severe enough to interfere with walking, running, and other activities of daily life. MYOSCN4A is a phenotypically highly variable myotonia aggravated by potassium loading, and often by cold. MYOSCN4A includes myotonia permanens and myotonia fluctuans. In myotonia permanens, the myotonia is generalized and there is a hypertrophy of the muscle, particularly in the neck and the shoulder. Attacks of severe muscle stiffness of the thoracic muscles may be life threatening due to impaired ventilation. In myotonia fluctuans, the muscle stiffness may

	fluctuate from day to day, provoked by exercise. Defects in SCN4A are the cause of a congenital myasthenic syndrome acetazolamide-responsive (CMSAR) [MIM:614198]. A congenital myasthenic syndrome associated with fatigable generalized weakness and recurrent attacks of respiratory and bulbar paralysis since birth. The fatigable weakness involves lid-elevator, external ocular, facial, limb and truncal muscles and an decremental response of the compound muscle action potential on repetitive stimulation.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	Voltage-gated sodium channels are transmembrane glycoprotein complexes composed of a large alpha subunit with 24 transmembrane domains and one or more regulatory beta subunits. They are responsible for the generation and propagation of action potentials in neurons and muscle. This gene encodes one member of the sodium channel alpha subunit gene family. It is expressed in skeletal muscle, and mutations in this gene have been linked to several myotonia and periodic paralysis disorders. [provided by RefSeq, Jul 2008]

Additional Information

Gene ID	6329
Other Names	Sodium channel protein type 4 subunit alpha, SkM1, Sodium channel protein skeletal muscle subunit alpha, Sodium channel protein type IV subunit alpha, Voltage-gated sodium channel subunit alpha Nav1.4, SCN4A (HGNC:10591)
Dilution	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	SCN4A (HGNC:10591)
Function	Pore-forming subunit of Nav1.4, a voltage-gated sodium (Nav) channel that directly mediates the depolarizing phase of action potentials in excitable membranes. Navs, also called VGSCs (voltage- gated sodium channels) or VDSCs (voltage-dependent sodium channels), operate by switching between closed and open conformations depending on the voltage difference across the membrane. In the open conformation they allow Na(+) ions to selectively pass through the pore, along their electrochemical gradient. The influx of Na+ ions provokes membrane depolarization, initiating the propagation of electrical signals throughout cells and tissues (PubMed:12766226, PubMed:15318338, PubMed:16890191, PubMed:17898326, PubMed:18690054, PubMed:19347921, PubMed:25707578, PubMed:26659129, PubMed:26700687, PubMed:29992740, PubMed:30190309). Highly expressed in skeletal muscles, Nav1.4 generates the action potential crucial for muscle contraction (PubMed:16890191, PubMed:19347921, PubMed:25707578, PubMed:26659129, PubMed:26700687).
Cellular Location	Cell membrane; Multi-pass membrane protein

Background

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