电压门控钠离子通道型α4抗体

产品货号： mlR17291

英文名称： SCN4A

中文名称： 电压门控钠离子通道型α4抗体

别　　名： HYKPP; HYPP; Na(V)1.4; NAC1A; Nav1.4; Scn4a; SCN4A_HUMAN; Skeletal muscle voltage dependent sodium channel type IV alpha subunit; SkM1; Sodium channel protein skeletal muscle subunit alpha; Sodium channel protein type 4 subunit alpha; Sodium channel protein type IV subunit alpha; Sodium channel voltage gated type IV alpha subunit; Voltage gated sodium channel subunit alpha Nav1.4; Voltage gated sodium channel type 4 alpha; Voltage-gated sodium channel subunit alpha Nav1.4.

研究领域： 免疫学　神经生物学　通道蛋白　跨膜蛋白

抗体来源： Rabbit

克隆类型： Polyclonal

交叉反应： Human, Mouse, Rat, Cow, Sheep,

产品应用 ： ELISA=1:500-1000 IHC-P=1:400-800 IHC-F=1:400-800 ICC=1:100-500 IF=1:100-500 （石蜡切片需做抗原修复）

not yet tested in other applications.

optimal dilutions/concentrations should be determined by the end user.

分 子 量 ： 208kDa

细胞定位 ： 细胞膜

性　　状 ： Lyophilized or Liquid

浓　　度 ： 1mg/ml

免 疫 原 ： KLH conjugated synthetic peptide derived from human SCN4A:301-400/1836 <Extracellular>

亚　　型 ： IgG

纯化方法 ： affinity purified by Protein A

储 存 液 ： 0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.

保存条件 ： 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.

PubMed : PubMed

产品介绍 : 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]

Function:

This protein mediates the voltage-dependent sodium ion permeability of excitable membranes. Assuming opened or closed conformations in response to the voltage difference across the membrane, the protein forms a sodium-selective channel through which Na(+) ions may pass in accordance with their electrochemical gradient. This sodium channel may be present in both denervated and innervated skeletal muscle.

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.

Similarity:

Belongs to the sodium channel (TC 1.A.1.10) family. Nav1.4/SCN4A subfamily. Contains 1 IQ domain.

SWISS:

P35499

Gene ID:

6329

Important Note:

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