abcam

Product datasheet

Recombinant Human SCN2A protein ab114727

画像数 1

製品の詳細

製品名	Recombinant Human SCN2A protein
発現系	Wheat germ
アクセッション番号	Q99250
タンパク質長	Protein fragment
Animal free	No
由来	Recombinant
生物種	Human
配列	NLRNKCLQWPPDNSSFEINITSFFNNSLDGNGTTFNRTVSIF NWDEYIED KSHFYFLEGQNDALLCGNSSDAGQCPEGYICVKAGRNPNY
予測される分子量	36 kDa including tags
領域	273 to 362

特性

Our **Abpromise guarantee** covers the use of **ab114727** in the following tested applications.

The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

アプリケーション	ELISA SDS-PAGE Western blot
製品の状態	Liquid

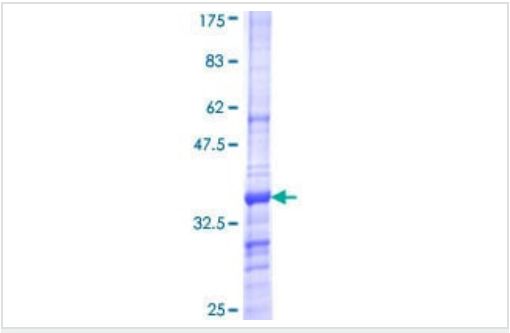
前処理および保存

保存方法および安定性	Shipped on dry ice. Upon delivery aliquot and store at -80ºC. Avoid freeze / thaw cycles. pH: 8.00 Constituents: 0.3% Glutathione, 0.79% Tris HCl

関連情報

機能	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.
関連疾患	Defects in SCN2A are a cause of generalized epilepsy with febrile seizures plus (GEFS+) [MIM:604233]. Generalized epilepsy with febrile seizures-plus refers to a rare autosomal dominant, familial condition with incomplete penetrance and large intrafamilial variability. Patients display febrile seizures persisting sometimes beyond the age of 6 years and/or a variety of afebrile seizure types. GEFS+ is a disease combining febrile seizures, generalized seizures often precipitated by fever at age 6 years or more, and partial seizures, with a variable degree of severity. Defects in SCN2A are the cause of benign familial infantile convulsions type 3 (BFIC3) [MIM:607745]. BFIC3 is an autosomal dominant disorder in which afebrile seizures occur in clusters during the first year of life, without neurologic sequelae. Defects in SCN2A are the cause of epileptic encephalopathy early infantile type 11 (EIEE11) [MIM:613721]. EIEE11 is an autosomal dominant seizure disorder characterized by infantile onset of refractory seizures with resultant delayed neurologic development and persistent neurologic abnormalities.
配列類似性	Belongs to the sodium channel (TC 1.A.1.10) family. Nav1.2/SCN2A subfamily. Contains 1 IQ domain.
ドメイン	The sequence contains 4 internal repeats, each with 5 hydrophobic segments (S1,S2,S3,S5,S6) and one positively charged segment (S4). Segments S4 are probably the voltage-sensors and are characterized by a series of positively charged amino acids at every third position.
翻訳後修飾	May be ubiquitinated by NEDD4L; which would promote its endocytosis.
細胞内局在	Membrane.

画像



SDS-PAGE - Recombinant Human SCN2A protein (ab114727)

12.5% SDS-PAGE Stained with Coomassie Blue

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