abcam

Product datasheet

Recombinant Human SCN2A protein ab114727

1 Image

Description

Product name	Recombinant Human SCN2A protein
Expression system	Wheat germ
Accession	**Q99250**
Protein length	Protein fragment
Animal free	No
Nature	Recombinant
Species	Human
Sequence	NLRNKCLQWPPDNSSFEINITSFFNNSLDGNGTTFNRTVSIFNWDEYIED KSHFYFLEGQNDALLCGNSSDAGQCPEGYICVKAGRNPNY
Predicted molecular weight	36 kDa including tags
Amino acids	273 to 362

Specifications

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.

Applications	ELISA SDS-PAGE Western blot
Form	Liquid

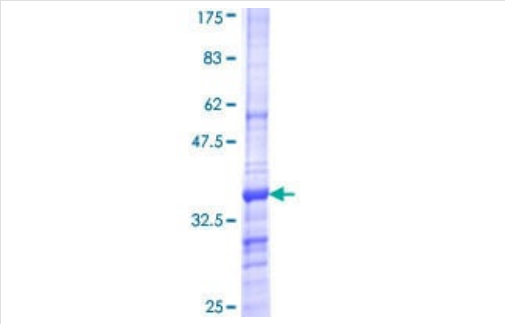
Preparation and Storage

Stability and Storage	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

General Info

Function	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.
Involvement in disease	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.
Sequence similarities	Belongs to the sodium channel (TC 1.A.1.10) family. Nav1.2/SCN2A subfamily. Contains 1 IQ domain.
Domain	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.
Post-translational modifications	May be ubiquitinated by NEDD4L; which would promote its endocytosis.
Cellular localization	Membrane.

Images



SDS-PAGE - Recombinant Human SCN2A protein (ab114727)

12.5% SDS-PAGE Stained with Coomassie Blue

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