abcam

Product datasheet

Recombinant Human Rapsyn protein ab114714

画像数 1

製品の概要

製品名	Recombinant Human Rapsyn protein
タンパク質長	Protein fragment

製品の詳細

由来	Recombinant
由来	Wheat germ
アミノ酸配列	
アクセッション番号	Q13702
生物種	Human
配列	QDLAEEVGNKLSQLKLHCLSESIYRSKGLQRELRAHVVRFHECVEETELY CGLCGESIGEKNSRLQALPCSHIFHLRCLQNNGTRSCPNCRRSSMKPGFV
分子量	37 kDa including tags
領域	313 to 412

特性

Our Abpromise guarantee covers the use of **ab114714** 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
備考	Protein concentration is above or equal to 0.05 mg/ml. Best use within three months from the date of receipt of this protein.

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

Thought to play some role in anchoring or stabilizing the nicotinic acetylcholine receptor at synaptic sites. It may link the receptor to the underlying postsynaptic cytoskeleton, possibly by direct association with actin or spectrin.

関連疾患

Defects in RAPSN are a cause of congenital myasthenic syndrome with acetylcholine receptor deficiency (ACHRDCMS) [MIM:608931]. ACHRDCMS is a post-synaptic congenital myasthenic syndrome. Congenital myasthenic syndromes (CMS) are inherited disorders of neuromuscular transmission that stem from mutations in presynaptic, synaptic, or postsynaptic proteins. Postsynaptic disorders result from mutations in proteins forming the subunits of the muscle acetylcholine receptor (AChR). The kinetic abnormalities of AChR result in either prolonged ion channel activations that underlie 'slow-channel myasthenic syndromes' (SCCMS) or abbreviated channel activations that underlie the abnormally rapid decay of endplate currents in 'fast-channel syndromes' (FCCMS). ACHRDCMS is the third disorder associated with postsynaptic CMS which could result from mutations in the proteins forming the muscle AChR. Mutations underlying AChR deficiency cause a 'loss of function' and show recessive inheritance.
Defects in RAPSN are the cause of fetal akinesia deformation sequence (FADS) [MIM:208150]; also known as Pena-Shokeir syndrome type 1 or fetal akinesia sequence or arthrogryposis multiplex congenita with pulmonary hypoplasia. FADS is a rare condition characterized by decreased intrauterine fetal movement, congenital limb contractures, pulmonary hypoplasia, polyhydramnios and craniofacial abnormalities.

配列類似性

Belongs to the RAPsyn family.
Contains 1 RING-type zinc finger.
Contains 7 TPR repeats.

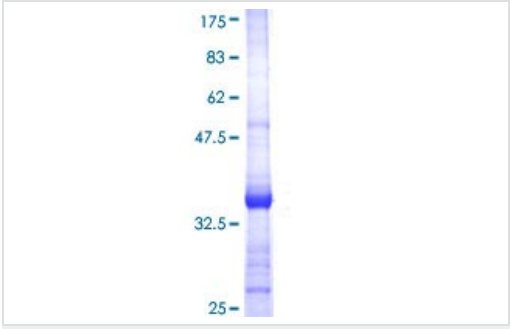
ドメイン

A cysteine-rich region homologous to part of the regulatory domain of protein kinase C may be important in interactions of this protein with the lipid bilayer.

細胞内局在

Cell membrane. Cell junction > synapse > postsynaptic cell membrane. Cytoplasm > cytoskeleton. Cytoplasmic surface of postsynaptic membranes.

画像



SDS-PAGE - Rapsyn protein (ab114714)

12.5% SDS-PAGE showing ab114714 at approximately 36.63 kDa stained with Coomassie Blue.

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