abcam

Product datasheet

Recombinant Human Gephyrin protein ab153064

1 Image

Overview

Product name	Recombinant Human Gephyrin protein
Protein length	Protein fragment

Description

Nature	Recombinant
Source	Wheat germ
Amino Acid Sequence	
Species	Human
Sequence	RKMQGILDPRPTIIKARLSCDVKLDPRPEYHRCILTWHH QEPLPWAQSTG NQMSSRLMSMRSANGLLMLPPKTEQYVELHKGEVVD VMVIGRL
Amino acids	677 to 769
Tags	proprietary tag N-Terminus

Specifications

Our Abpromise guarantee covers the use of **ab153064** in the following tested applications.

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

Applications	ELISA Western blot
Form	Liquid
Additional notes	Protein concentration is above or equal to 0.05 mg/ml.

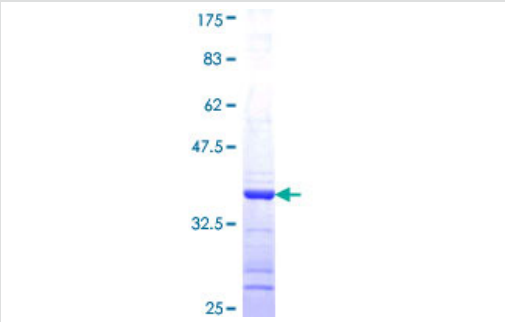
Preparation and Storage

Stability and Storage	Shipped on dry ice. Upon delivery aliquot and store at -80ºC. Avoid freeze / thaw cycles. pH: 8.00 Constituents: 0.31% Glutathione, 0.79% Tris HCl

General Info

Function	Microtubule-associated protein involved in membrane protein-cytoskeleton interactions. It is thought to anchor the inhibitory glycine receptor (GLYR) to subsynaptic microtubules (By similarity). Catalyzes two steps in the biosynthesis of the molybdenum cofactor. In the first step, molybdopterin is adenylated. Subsequently, molybdate is inserted into adenylated molybdopterin and AMP is released.
Pathway	Cofactor biosynthesis; molybdopterin biosynthesis.
Involvement in disease	Defects in GPHN are the cause of molybdenum cofactor deficiency type C (MOCOD type C) [MIM:252150]. MOCOD type C is an autosomal recessive disease which leads to the pleiotropic loss of all molybdoenzyme activities and is characterized by severe neurological damage, neonatal seizures and early childhood death. Defects in GPHN are a cause of startle disease (STHE) [MIM:149400]; also known as hyperekplexia. STHE is a genetically heterogeneous neurologic disorder characterized by muscular rigidity of central nervous system origin, particularly in the neonatal period, and by an exaggerated startle response to unexpected acoustic or tactile stimuli.
Sequence similarities	In the N-terminal section; belongs to the moaB/mog family. In the C-terminal section; belongs to the moeA family.
Cellular localization	Cell junction > synapse. Cell junction > synapse > postsynaptic cell membrane. Cytoplasm > cytoskeleton. Cytoplasmic face of glycinergic postsynaptic membranes.

Images



SDS-PAGE - Recombinant Human Gephyrin protein (ab153064)

ab153064 on a 12.5% SDS-PAGE stained with Coomassie Blue.

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