

Product datasheet

Recombinant Human Gephyrin protein ab153064

1 Image

Overview

<b>Product name</b>	Recombinant Human Gephyrin protein
<b>Protein length</b>	Protein fragment

Description

<b>Nature</b>	Recombinant
<b>Source</b>	Wheat germ

Amino Acid Sequence

<b>Species</b>	Human
<b>Sequence</b>	RKMQGILDPRTIHKARLSCDVKLDPRPEYHRCILTWHHQEPLPWAQSTG NQMSRLMSMRSANGLLMLPPKTEQYVELHKGEVVDVMVIGRL
<b>Amino acids</b>	677 to 769
<b>Tags</b>	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.

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

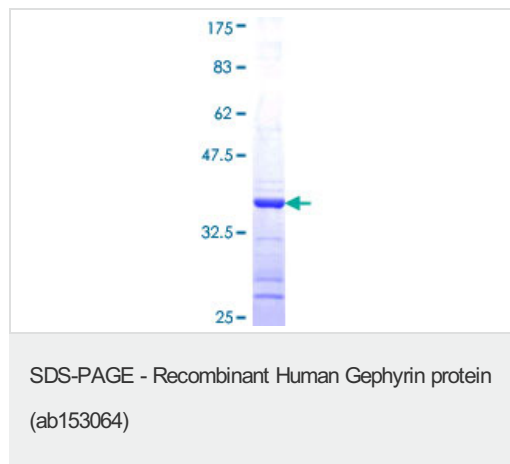
Preparation and Storage

<b>Stability and Storage</b>	Shipped on dry ice. Upon delivery aliquot and store at -80°C. Avoid freeze / thaw cycles. pH: 8.00 Constituents: 0.31% Glutathione, 0.79% Tris HCl
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General Info

<b>Function</b>	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.
<b>Pathway</b>	Cofactor biosynthesis; molybdopterin biosynthesis.
<b>Involvement in disease</b>	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.
<b>Sequence similarities</b>	In the N-terminal section; belongs to the moaB/mog family. In the C-terminal section; belongs to the moeA family.
<b>Cellular localization</b>	Cell junction > synapse. Cell junction > synapse > postsynaptic cell membrane. Cytoplasm > cytoskeleton. Cytoplasmic face of glycinergic postsynaptic membranes.

## Images



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

**Please note:** All products are "FOR RESEARCH USE ONLY AND ARE NOT INTENDED FOR DIAGNOSTIC OR THERAPEUTIC USE"

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