abcam

Product datasheet

Recombinant Human Connexin 32 / GJB1 protein ab158522

1 Image

Description

Product name Recombinant Human Connexin 32 / GJB1 protein

Expression system Wheat germ

Protein length Full length protein

Animal free No

Nature Recombinant

Species Human

Sequence MNWTGLYTLLSGVNRHSTAIGRVWLSVIFIFRIMVLVVAAES

VWGDEKSS

 ${\sf FICNTLQPGCNSVCYDQFFPISHVRLWSLQLILVSTPALLV}$

AMHVAHQQH

IEKKMLRLEGHGDPLHLEEVKRHKVHISGTLWWTYVISVVF

RLLFEAVFM

YVFYLLYPGYAMVRLVKCDVYPCPNTVDCFVSRPTEKTVF

TVFMLAASGI

CIILNVAEVVYLIIRACARRAQRRSNPPSRKGSGFGHRLSP

EYKQNEINK

LLSEQDGSLKDILRRSPGTGAGLAEKSDRCSAC

Amino acids 1 to 283

Tags GST tag N-Terminus

Specifications

Our Abpromise guarantee covers the use of ab158522 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

1

Preparation and Storage

Stability and Storage

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 HCI

General Info

Function

Involvement in disease

One gap junction consists of a cluster of closely packed pairs of transmembrane channels, the connexons, through which materials of low MW diffuse from one cell to a neighboring cell.

Defects in GJB1 are the cause of Charcot-Marie-Tooth disease X-linked type 1 (CMTX1) [MIM:302800]; also designated CMT-X. CMTX1 is a form of Charcot-Marie-Tooth disease, the most common inherited disorder of the peripheral nervous system. Charcot-Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies characterized by severely reduced motor nerve conduction velocities (NCVs) (less than 38m/s) and segmental demyelination and remyelination, and primary peripheral axonal neuropathies characterized by normal or mildly reduced NCVs and chronic axonal degeneration and regeneration on nerve biopsy. CMTX1 has both demyelinating and axonal features. Central nervous system involvement may occur.

Defects in GJB1 may contribute to the phenotype of Dejerine-Sottas syndrome (DSS) [MIM:145900]; also known as Dejerine-Sottas neuropathy (DSN) or hereditary motor and sensory neuropathy III (HMSN3). DSS is a severe degenerating neuropathy of the demyelinating Charcot-Marie-Tooth disease category, with onset by age 2 years. DSS is characterized by motor and sensory neuropathy with very slow nerve conduction velocities, increased cerebrospinal fluid protein concentrations, hypertrophic nerve changes, delayed age of walking as well as areflexia. There are both autosomal dominant and autosomal recessive forms of Dejerine-Sottas syndrome.

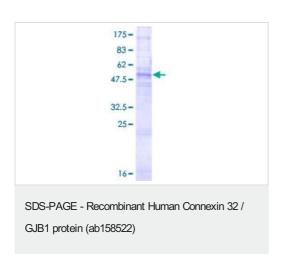
Sequence similarities

Cellular localization

Belongs to the connexin family. Beta-type (group I) subfamily.

Cell membrane. Cell junction > gap junction.

Images



ab158522 on a 12.5% SDS-PAGE stained with Coomassie Blue.

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