abcam

Product datasheet

Recombinant Human PMP22 protein ab112342

1 Image

Description

Product name	Recombinant Human PMP22 protein
Expression system	Wheat germ
Accession	Q01453
Protein length	Full length protein
Animal free	No
Nature	Recombinant
Species	Human
Sequence	MLLLLLSIIVLHVAVLVLLFVSTIVSQWIVGNGHATDLWQNC STSSSGNV HHCFSSSPNEWLQSVQATMILSIIFSILSLFLFFCQLFTLTK GGRFYITG IFQILAGLCVMSAAAIYTVRHPEWHLNSDYSYGFAYILAWVA FPLALLSG VIYVILRKRE
Predicted molecular weight	45 kDa including tags
Amino acids	1 to 160
Tags	GST tag N-Terminus

Specifications

Our **Abpromise guarantee** covers the use of **ab112342** in the following tested applications.

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

Applications	SDS-PAGE ELISA Western blot
Form	Liquid

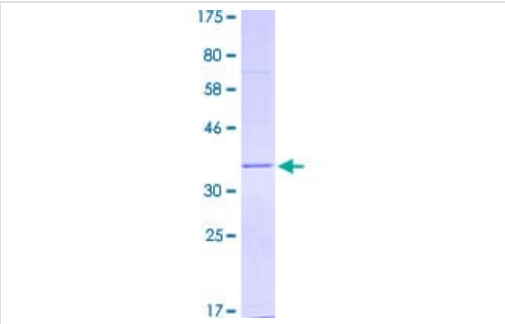
Preparation and Storage

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

General Info

Function	Might be involved in growth regulation, and in myelinization in the peripheral nervous system.
Involvement in disease	Defects in PMP22 are the cause of Charcot-Marie-Tooth disease type 1A (CMT1A) [MIM:118220]; also known as hereditary motor and sensory neuropathy IA. CMT1A 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 neuropathy or CMT1, and primary peripheral axonal neuropathy or CMT2. Neuropathies of the CMT1 group are characterized by severely reduced nerve conduction velocities (less than 38 m/sec), segmental demyelination and remyelination with onion bulb formations on nerve biopsy, slowly progressive distal muscle atrophy and weakness, absent deep tendon reflexes, and hollow feet. CMT1A inheritance is autosomal dominant. Defects in PMP22 are a cause 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. Defects in PMP22 are a cause of hereditary neuropathy with liability to pressure palsies (HNPP) [MIM:162500]; an autosomal dominant disorder characterized by transient episodes of decreased perception or peripheral nerve palsies after slight traction, compression or minor traumas. Defects in PMP22 are the cause of Charcot-Marie-Tooth disease type 1E (CMT1E) [MIM:118300]; also known as Charcot-Marie-Tooth disease and deafness autosomal dominant. CMT1E is an autosomal dominant form of Charcot-Marie-Tooth disease characterized by the association of sensorineural hearing loss with peripheral demyelinating neuropathy. Defects in PMP22 may be a cause of inflammatory demyelinating polyneuropathy (IDP) [MIM:139393]. IDP is a putative autoimmune disorder presenting in an acute (AIDP) or chronic form (CIDP). The acute form is also known as Guillain-Barre syndrome.
Sequence similarities	Belongs to the PMP-22/EMP/MP20 family.
Cellular localization	Membrane.

Images



SDS-PAGE - Recombinant Human PMP22 protein (ab112342)

ab112342 analysed on a 12.5% SDS-PAGE gel stained with Coomassie Blue.

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