

Product datasheet

Recombinant Human PMP22 protein ab112342

1 图像

描述

产品名称	重组人PMP22蛋白
表达系统	Wheat germ
Accession	<u>Q01453</u>
蛋白长度	Full length protein
无动物成分	No
性质	Recombinant
种属	Human
序列	MLLLLLSIIVLHVAVLVLLFVSTIVSQWIVGNGHATDLWQNC STSSSGNV HHCFS SSPNEWLQSVQATMILSIIIFSILSLFLFFCQLFTLTK GGRFYITG IFQILAGLCVMSAAAIIYTVRHPEWHLNSDYSYGFAYILAWVA FPLALLSG VIYVILRKRE
预测分子量	45 kDa including tags
氨基酸	1 to 160
标签	GST tag N-Terminus

技术指标

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.

应用	SDS-PAGE ELISA Western blot
形式	Liquid

制备和贮存

稳定性和存储	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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常规信息

功能

Might be involved in growth regulation, and in myelination in the peripheral nervous system.

疾病相关

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.

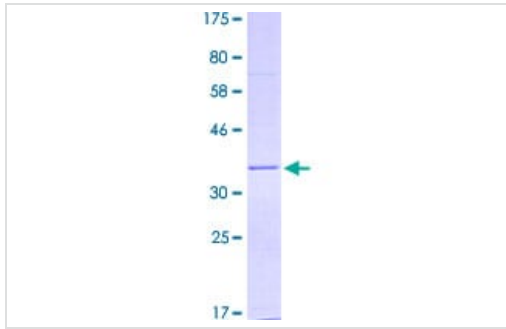
序列相似性

Belongs to the PMP-22/EMP/MP20 family.

细胞定位

Membrane.

图片



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

SDS-PAGE - Recombinant Human PMP22 protein (ab112342)

Please note: All products are "FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC PROCEDURES"

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