abcam

Product datasheet

Recombinant Human MPV17 protein ab116947

1 Image

Overview

Product name	Recombinant Human MPV17 protein
Protein length	Full length protein

Description

Nature	Recombinant
Source	Wheat germ

Amino Acid Sequence

Accession	P39210
Species	Human
Sequence	MALWRAYQRALAAHPWKVQVLTAGSLMGLGDIISQQLVERRGLQEHQRGR TLTMVSLGCGFVGPVVGGWYKVLDRFIPGTTKVDALKKMLLDQGGFAPCF LGCFLPLVGALNGLSAQDNWAKLQRDYPDALITNYYLWPAVQLANFYLVP LHYRLAVVQCVAVIWNSYLSWKAHRL
Molecular weight	45 kDa including tags
Amino acids	1 to 176

Specifications

Our Abpromise guarantee covers the use of **ab116947** in the following tested applications.

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

Applications	ELISA SDS-PAGE Western blot
Form	Liquid
Additional notes	Protein concentration is above or equal to 0.05 mg/ml. Best used within three months from the date of receipt.

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: 0.3% Glutathione, 0.79% Tris HCl

General Info

Function
Involved in mitochondria homeostasis. May be involved in the metabolism of reactive oxygen species and control of oxidative phosphorylation and mitochondrial DNA (mtDNA) maintenance.

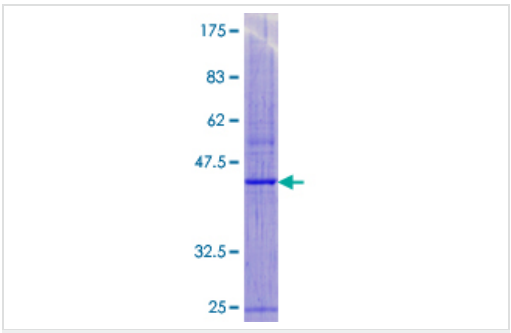
Tissue specificity
Ubiquitous. Expressed in pancreas, kidney, muscle, liver, lung, placenta, brain and heart.

Involvement in disease
Defects in MPV17 are a cause of hepatocerebral mitochondrial DNA depletion syndrome (MDS) [MIM:251880]. MDS is a clinically heterogeneous group of disorders characterized by a reduction in mitochondrial DNA (mtDNA) copy number. Primary mtDNA depletion is inherited as an autosomal recessive trait and may affect single organs, typically muscle or liver, or multiple tissues. Individuals with the hepatocerebral form of mitochondrial DNA depletion syndrome have early progressive liver failure and neurologic abnormalities, hypoglycemia, and increased lactate in body fluids.
Defects in MPV17 are the cause of Navajo neurohepatopathy (NN) [MIM:256810]. NN is an autosomal recessive disease that is prevalent among Navajo children in the southwestern United States. The major clinical features are hepatopathy, peripheral neuropathy, corneal anesthesia and scarring, acral mutilation, cerebral leukoencephalopathy, failure to thrive, and recurrent metabolic acidosis with intercurrent infections. Infantile, childhood, and classic forms of NN have been described. Mitochondrial DNA depletion was detected in the livers of patients, suggesting a primary defect in mtDNA maintenance.

Sequence similarities
Belongs to the peroxisomal membrane protein PXMP2/4 family.

Cellular localization
Mitochondrion inner membrane.

Images



SDS-PAGE - MPV17 protein (ab116947)

12.5% SDS-PAGE showing ab116947 at approximately 45.53kDa stained with Coomassie Blue.

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