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Product datasheet

Recombinant Human ATP7A protein ab114343

1 Image

Overview

Product name	Recombinant Human ATP7A protein
Protein length	Protein fragment

Description

Nature	Recombinant
Source	Wheat germ

Amino Acid Sequence

Accession	Q04656
Species	Human
Sequence	FLKLYRKPTYESYELPARSQIGQKSPSEISVHVGIDDTSRNSPKLGLLDR IVNYSRASINSLLSDKRSLNSVVTSEPDKHSLLVGDFREDDDTAL
Molecular weight	36 kDa including tags
Amino acids	1406 to 1500

Specifications

Our Abpromise guarantee covers the use of **ab114343** 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 use within three months from the date of receipt of this protein.

Preparation and Storage

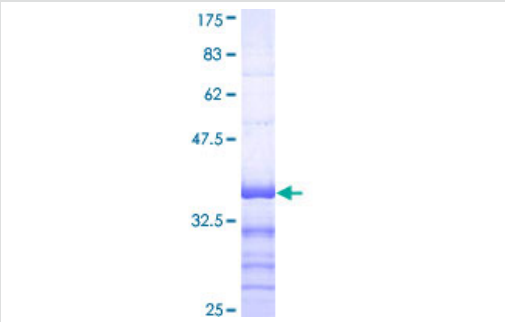
Stability and Storage	Shipped on dry ice. Upon delivery aliquot and store at -80ºC. Avoid freeze / thaw cycles. pH: 8.00

Constituents: 0.3% Glutathione, 0.79% Tris HCl

General Info

Function	May supply copper to copper-requiring proteins within the secretory pathway, when localized in the trans-Golgi network. Under conditions of elevated extracellular copper, it relocalized to the plasma membrane where it functions in the efflux of copper from cells.
Tissue specificity	Found in most tissues except liver. Isoform 3 is widely expressed including in liver cell lines. Isoform 1 is expressed in fibroblasts, choriocarcinoma, colon carcinoma and neuroblastoma cell lines. Isoform 2 is expressed in fibroblasts, colon carcinoma and neuroblastoma cell lines.
Involvement in disease	Defects in ATP7A are the cause of Menkes disease (MNKD) [MIM:309400]; also known as kinky hair disease. MNKD is an X-linked recessive disorder of copper metabolism characterized by generalized copper deficiency. MNKD results in progressive neurodegeneration and connective-tissue disturbances: focal cerebral and cerebellar degeneration, early growth retardation, peculiar hair, hypopigmentation, cutis laxa, vascular complications and death in early childhood. The clinical features result from the dysfunction of several copper-dependent enzymes. Defects in ATP7A are the cause of occipital horn syndrome (OHS) [MIM:304150]; also known as X-linked cutis laxa. OHS is an X-linked recessive disorder of copper metabolism. Common features are unusual facial appearance, skeletal abnormalities, chronic diarrhea and genitourinary defects. The skeletal abnormalities included occipital horns, short, broad clavicles, deformed radii, ulnae and humeri, narrowing of the rib cage, undercalcified long bones with thin cortical walls and coxa valga. Defects in ATP7A are a cause of distal spinal muscular atrophy X-linked type 3 (DSMAX3) [MIM:300489]. DSMAX3 is a neuromuscular disorder. Distal spinal muscular atrophy, also known as distal hereditary motor neuronopathy, represents a heterogeneous group of neuromuscular disorders caused by selective degeneration of motor neurons in the anterior horn of the spinal cord, without sensory deficit in the posterior horn. The overall clinical picture consists of a classical distal muscular atrophy syndrome in the legs without clinical sensory loss. The disease starts with weakness and wasting of distal muscles of the anterior tibial and peroneal compartments of the legs. Later on, weakness and atrophy may expand to the proximal muscles of the lower limbs and/or to the distal upper limbs.
Sequence similarities	Belongs to the cation transport ATPase (P-type) (TC 3.A.3) family. Type IB subfamily. Contains 6 HMA domains.
Domain	The C-terminal di-leucine, 1487-Leu-Leu-1488, is an endocytic targeting signal which functions in retrieving recycling from the plasma membrane to the TGN. Mutation of the di-leucine signal results in the accumulation of the protein in the plasma membrane.
Cellular localization	Endoplasmic reticulum; Cytoplasm > cytosol and Golgi apparatus > trans-Golgi network membrane. Cell membrane. Cycles constitutively between the trans-Golgi network (TGN) and the plasma membrane. Predominantly found in the TGN and relocalized to the plasma membrane in response to elevated copper levels.

Images



SDS-PAGE - ATP7A protein (ab114343)

ab114343 analysed on a 12.5% SDS-PAGE gel stained with Coomassie Blue.

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