abcam

Product datasheet

Recombinant Human Frataxin protein ab95502

1 Image

Description

Product name	Recombinant Human Frataxin protein
Purity	> 95 % SDS-PAGE. ab95502 is purified using conventional chromatography techniques.
Expression system	Escherichia coli
Accession	Q16595
Protein length	Full length protein
Animal free	No
Nature	Recombinant
Species	Human
Sequence	**MGSSHHHHHH SSGLVPRGSH** MLRTDIDATC TPRRASSNQR GLNQIWNVKK QSVYLMNLRK SGTLGHPGSL DETTYERLAE ETLDSLAEFF EDLADKPYTF EDYDVSFGSG VLTVKLGGDL GTYVINKQTP NKQIWLSSPS SGPKRYDWTG KNWVYSHDGV SLHELLAAEL TKALKTKLDL SSLAYSGKDA
Predicted molecular weight	21 kDa including tags
Tags	His tag N-Terminus

Specifications

Our Abpromise guarantee covers the use of **ab95502** 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 Mass Spectrometry
Form	Liquid

Preparation and Storage

Stability and Storage	Shipped at 4°C. Upon delivery aliquot and store at -20°C or -80°C. Avoid repeated freeze / thaw cycles.

pH: 8.00
Constituents: 0.0154% DTT, 0.316% Tris HCl, 10% Glycerol

General Info

Function

Promotes the biosynthesis of heme and assembly and repair of iron-sulfur clusters by delivering Fe(2+) to proteins involved in these pathways. May play a role in the protection against iron-catalyzed oxidative stress through its ability to catalyze the oxidation of Fe(2+) to Fe(3+); the oligomeric form but not the monomeric form has in vitro ferroxidase activity. May be able to store large amounts of iron in the form of a ferrihydrite mineral by oligomerization; however, the physiological relevance is unsure as reports are conflicting and the function has only been shown using heterologous overexpression systems. Modulates the RNA-binding activity of ACO1.

Tissue specificity

Expressed in the heart, peripheral blood lymphocytes and dermal fibroblasts.

Involvement in disease

Defects in FXN are the cause of Friedreich ataxia (FRDA) [MIM:229300]. FRDA is an autosomal recessive, progressive degenerative disease characterized by neurodegeneration and cardiomyopathy it is the most common inherited ataxia. The disorder is usually manifest before adolescence and is generally characterized by incoordination of limb movements, dysarthria, nystagmus, diminished or absent tendon reflexes, Babinski sign, impairment of position and vibratory senses, scoliosis, pes cavus, and hammer toe. In most patients, FRDA is due to GAA triplet repeat expansions in the first intron of the frataxin gene. But in some cases the disease is due to mutations in the coding region.

Sequence similarities

Belongs to the frataxin family.

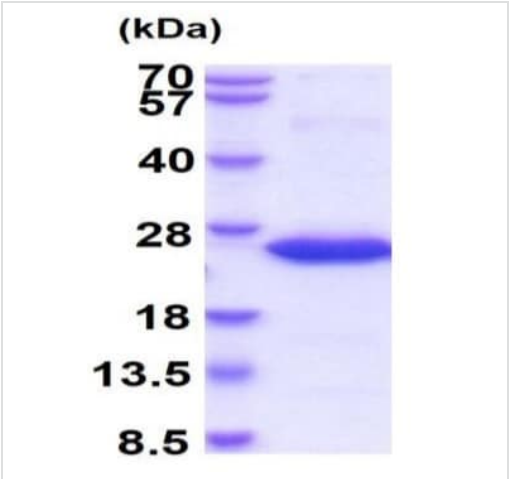
Post-translational modifications

Processed in two steps by mitochondrial processing peptidase (MPP). MPP first cleaves the precursor to intermediate form and subsequently converts the intermediate to yield frataxin mature form (frataxin(81-210)) which is the predominant form. The additional forms, frataxin(56-210) and frataxin(78-210), seem to be produced when the normal maturation process is impaired; their physiological relevance is unsure.

Cellular localization

Cytoplasm. Mitochondrion. PubMed:18725397 reports localization exclusively in mitochondria.

Images



SDS-PAGE - Recombinant Human Frataxin protein (ab95502)

15% SDS-PAGE analysis of 3µg ab95502.

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

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