abcam

Product datasheet

Recombinant Human Aprataxin protein ab40478

1 Image

Overview

Product name	Recombinant Human Aprataxin protein
Protein length	Full length protein

Description

Nature	Recombinant
Source	Escherichia coli

Amino Acid Sequence

Accession	Q7Z2E3
Species	Human
Tags	His-T7 tag N-Terminus

Specifications

Our Abpromise guarantee covers the use of **ab40478** 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
Purity	> 90 % SDS-PAGE.
Form	Liquid

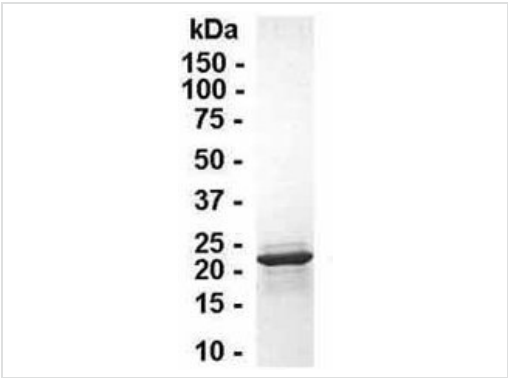
Preparation and Storage

Stability and Storage	Shipped at 4°C. Upon delivery aliquot. Store at -80°C. Avoid freeze / thaw cycle. Preservative: 0.002% Sodium Azide Constituents: 0.1% Triton-X-100, 10mM Tris, 10mM DTT, pH 8.0

General Info

Function	DNA-binding protein involved in single-strand DNA break repair, double-strand DNA break repair and base excision repair. Resolves abortive DNA ligation intermediates formed either at base excision sites, or when DNA ligases attempt to repair non-ligatable breaks induced by reactive oxygen species. Catalyzes the release of adenylate groups covalently linked to 5'-phosphate termini, resulting in the production of 5'-phosphate termini that can be efficiently rejoined. Also able to hydrolyze adenosine 5'-monophosphoramidate (AMP-NH(2)) and diadenosine tetraphosphate (AppppA), but with lower catalytic activity.
Tissue specificity	Widely expressed. In brain, it is expressed in the posterior cortex, cerebellum, hippocampus and olfactory bulb. Isoform 1 is highly expressed in the cerebral cortex and cerebellum, compared to isoform 2.
Involvement in disease	Defects in APTX are the cause of ataxia-oculomotor apraxia syndrome (AOA) [MIM:208920]. AOA is an autosomal recessive syndrome characterized by early-onset cerebellar ataxia, oculomotor apraxia, early areflexia and late peripheral neuropathy. Defects in APTX are a cause of coenzyme Q10 deficiency (COQ10D) [MIM:607426]. Coenzyme Q10 deficiency is an autosomal recessive disorder with variable manifestations. It can be associated with three main clinical phenotypes: a predominantly myopathic form with central nervous system involvement, an infantile encephalomyopathy with renal dysfunction and an ataxic form with cerebellar atrophy.
Sequence similarities	Contains 1 C2H2-type zinc finger. Contains 1 FHA-like domain. Contains 1 HIT domain.
Domain	The histidine triad, also called HIT motif, forms part of the binding loop for the alpha-phosphate of purine mononucleotide. The FHA-like domain mediates interaction with NCL; XRCC1 and XRCC4. The HIT domain is required for enzymatic activity. The C2H2-type zinc finger mediates DNA-binding.
Cellular localization	Nucleus > nucleoplasm. Nucleus > nucleolus. Upon genotoxic stress, colocalizes with XRCC1 at sites of DNA damage. Colocalizes with MDC1 at sites of DNA double-strand breaks. Interaction with NCL is required for nucleolar localization.

Images



SDS-PAGE - Aprataxin protein (T7 Tag) (ab40478)

4-20% SDS gradient gel. Coomassie blue staining.

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