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Product datasheet

Recombinant Human Aprataxin protein ab93630

1 Image

Overview

Product name	Recombinant Human Aprataxin protein
Protein length	Full length protein

Description

Nature	Recombinant
Source	Escherichia coli

Amino Acid Sequence

Species	Human
Sequence	MRGSHHHHHH GMASMTGGQQ MGRDLYDDDD KDRWAGSMQD PKMQVYKDEQ VVVIKDKYPK ARYHWLVLPW TSISSLKAVA REHLELLKHM HTVGEKVIVD FAGSSKLRFR LGYHAIPSMS HVHLHVISQD FDSPCLKNKK HWNSFNTEYF LESQAVIEMV QEAGRVTVRD GMPELLKLPL RCHECQQLLP SIPQLKEHLR KHWTQ
Amino acids	1 to 168

Specifications

Our Abpromise guarantee covers the use of **ab93630** 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
Purity	> 95 % SDS-PAGE. ab93630 is purified using conventional chromatography techniques.
Form	Liquid

Preparation and Storage

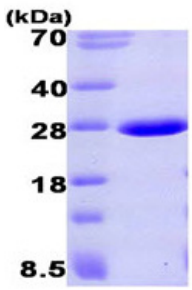
Stability and Storage	Shipped at 4°C. Store at +4°C short term (1-2 weeks). Upon delivery aliquot. Store at -20°C or -80°C. Avoid freeze / thaw cycle. Preservative: None

Constituents: 20% Glycerol, 20mM Tris HCl, 100mM Sodium chloride, 0.1mM PMSF, pH 7.5

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 (His tag) (ab93630)

15% SDS-PAGE showing ab93630 at approximately 23.9kDa (3µg).

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