abcam

Product datasheet

Recombinant Human Aprataxin protein ab93630

画像数1

製品の詳細

製品名 Recombinant Human Aprataxin protein

精製度 > 95 % SDS-PAGE.

ab93630 is purified using conventional chromatography techniques.

発現系 Escherichia coli

タンパク質長 Full length protein

Animal free No

由来 Recombinant

生物種 Human

配列 MRGSHHHHHH GMASMTGGQQ MGRDLYDDDD

KDRWAGSMQD PKMQVYKDEQ VVVIKDKYPK
ARYHWLVLPW TSISSLKAVA REHLELLKHM
HTVGEKVIVD FAGSSKLRFR LGYHAIPSMS
HVHLHVISQD FDSPCLKNKK HWNSFNTEYF
LESQAVIEMV QEAGRVTVRD GMPELLKLPL

RCHECQQLLP SIPQLKEHLR KHWTQ

領域 1 to 168

特性

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.

アプリケーション SDS-PAGE

製品の状態 Liquid

前処理および保存

保存方法および安定性 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.

pH: 7.50

Constituents: 0.00174% PMSF, 0.316% Tris HCl, 20% Glycerol (glycerin, glycerine), 0.58%

Sodium chloride

1

関連情報

機能

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.

組織特異性

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.

関連疾患

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.

配列類似性

Contains 1 C2H2-type zinc finger.
Contains 1 FHA-like domain.
Contains 1 HIT domain.

ドメイン

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.

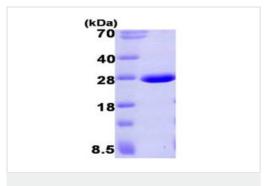
The HIT domain is required for enzymatic activity.

The C2H2-type zinc finger mediates DNA-binding.

細胞内局在

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.

画像



SDS-PAGE - Recombinant Human Aprataxin protein (ab93630)

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

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