abcam

Product datasheet

Recombinant Human Aprataxin protein ab93630

1 图像

描述

产品名称	重组人Aprataxin蛋白
纯度	> 95 % SDS-PAGE. ab93630 is purified using conventional chromatography techniques.
表达系统	Escherichia coli
蛋白长度	Full length protein
无动物成分	No
性质	Recombinant
种属	Human
序列	MRGSHHHHHH GMASMTGGQQ MGRDLYDDDD KDRWAGSMQD PKMQVYKDEQ VVVIKDKYPK ARYHWLVLPW TSISSLKAVA REHLELLKHM HTVGEKVIVD FAGSSKLRFR LGYHAIPSMS HVHLHVISQD FDSPCLKNKK HWNSFNTEYF LESQAVIEMV QEAGRVTVRD GMPELLKLPL RCHECQQLLP SIPQLKEHLR KHWTQ
氨基酸	1 to 168

技术指标

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.

应用	SDS-PAGE
形式	Liquid

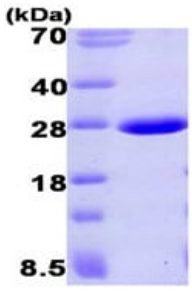
制备和贮存

稳定性和存储	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

常规信息

功能	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.
组织特异性	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.
疾病相关	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.
序列相似性	Contains 1 C2H2-type zinc finger. Contains 1 FHA-like domain. Contains 1 HIT domain.
结构域	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.
细胞定位	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.

图片



SDS-PAGE - Recombinant Human Aprataxin protein (ab93630)

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

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