



APT_X Polyclonal Antibody

Catalog No	YP-Ab-07349
Isotype	IgG
Reactivity	Human;Mouse;Rat
Applications	WB;ELISA
Gene Name	APT _X AXA1
Immunogen	Synthesized peptide derived from human protein . at AA range: 11-60
Specificity	APT _X Polyclonal Antibody detects endogenous levels of protein.
Formulation	Liquid in PBS containing 50% glycerol, and 0.02% sodium azide.
Source	Polyclonal, Rabbit,IgG
Purification	The antibody was affinity-purified from rabbit antiserum by affinity-chromatography using epitope-specific immunogen.
Dilution	WB 1:500-2000 ELISA 1:5000-20000
Concentration	1 mg/ml
Purity	≥90%
Storage Stability	-20°C/1 year
Synonyms	Forkhead-associated domain histidine triad-like protein;FHA-HIT;EC:3.6.1.71; AXA1;AXA 1;APT _X ;APT _X AXA1;Aprataxin (EC 3.-.-.-) (Forkhead-associated domain histidine triad-like protein) (FHA-HIT)
Observed Band	39kD
Calculated Molecular Weight	39kD
Cell Pathway	Nucleus, nucleoplasm . Nucleus, nucleolus . Upon genotoxic stress, colocalizes with XRCC1 at sites of DNA damage (PubMed:15380105). Colocalizes with MDC1 at sites of DNA double-strand breaks (PubMed:20008512). Interaction with NCL is required for nucleolar localization (PubMed:16777843). .; [Isoform 12]: Cytoplasm .
Tissue Specificity	Widely expressed; detected in liver, kidney and lymph node (at protein level) (PubMed:14755728). Isoform 1 is highly expressed in the cerebral cortex and cerebellum, compared to isoform 2 (at protein level) (PubMed:14755728). Widely expressed; detected throughout the brain, in liver, kidney, skeletal muscle, fibroblasts, lymphocytes and pancreas (PubMed:15276230, PubMed:11586299, PubMed:11586300).



Function

disease: Defects in APTX are a cause of coenzyme Q10 deficiency [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. Coenzyme Q10 deficiency due to APTX mutations is typically associated with cerebellar ataxia. 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. domain: The C2H2-type zinc finger mediates DNA-binding. domain: The FHA-like domain mediates interaction with NCL; XRCC1 and XRCC4. domain: The histidine triad, als

Background

aprataxin (APTX) Homo sapiens This gene encodes a member of the histidine triad (HIT) superfamily. The encoded protein may play a role in single-stranded DNA repair through its nucleotide-binding activity and its diadenosine polyphosphate hydrolase activity. Mutations in this gene have been associated with ataxia-ocular apraxia. Alternatively spliced transcript variants have been identified for this gene. [provided by RefSeq, Aug 2010],

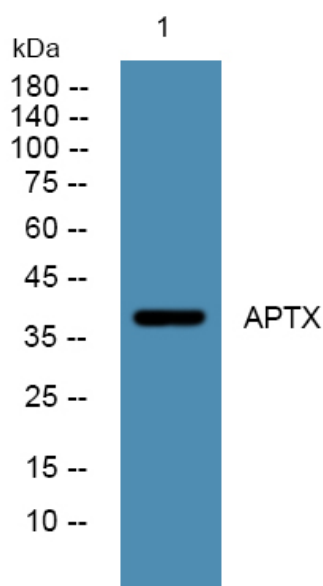
matters needing attention

Avoid repeated freezing and thawing!

Usage suggestions

This product can be used in immunological reaction related experiments. For more information, please consult technical personnel.

Products Images



Western blot analysis of lysates from HCT116 cells, primary antibody was diluted at 1:1000, 4° over night