



## **APTX Monoclonal Antibody**

Catalog No	BYmab-07349
Isotype	IgG
Reactivity	Human;Mouse;Rat
Applications	WB
Gene Name	APTX AXA1
Protein Name	Aprataxin (EC 3) (Forkhead-associated domain histidine triad-like protein) (FHA-HIT)
Immunogen	Synthesized peptide derived from human protein . at AA range: 11-60
Specificity	APTX Monoclonal Antibody detects endogenous levels of protein.
Formulation	Liquid in PBS containing 50% glycerol, and 0.02% sodium azide.
Source	Monoclonal, Mouse,lgG
Purification	The antibody was affinity-purified from mouse antiserum by affinity-chromatography using epitope-specific immunogen.
Dilution	WB 1:500-2000
Concentration	1 mg/ml
Purity	≥90%
Storage Stability	-20°C/1 year
Synonyms	
Observed Band	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

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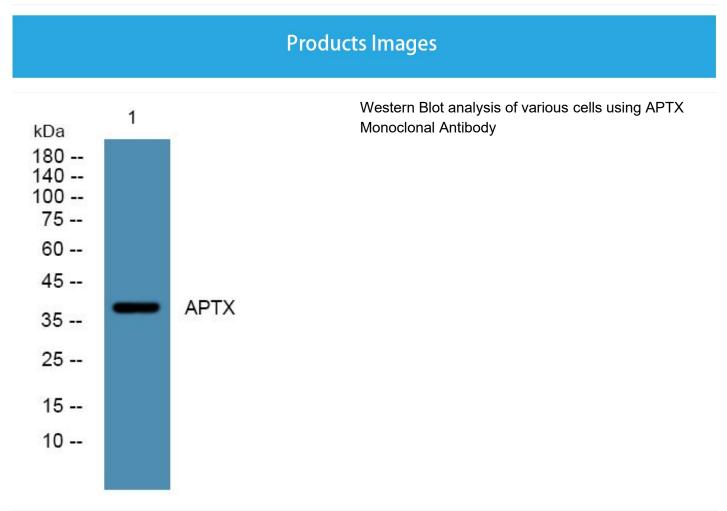
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	infantile encephalomyopathy with renal dysfunction and an ataxic form with cerebellar atrophy. Coenzyme Q10 deficiency due to APTX mutations is typically associated with cerebellar ataxia., disease:Defects in APTX are the cause of ataxia-oculomotor 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.



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