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Rabbit Anti-Aprataxin antibody

SL10524R

Product Name:	Aprataxin
Chinese Name:	共济失调性眼球运动功能丧失相关蛋白AOA1抗体
Alias:	AOA 1; AOA; AOA; AOA1; AOA1; Aprataxin; Aprataxin; Aprataxin homolog; APTX; APTX; APTX_HUMAN; Ataxia 1 early onset with hypoalbuminemia; Ataxia 1 early onset with hypoalbuminemia; Ataxia1 early onset with hypoalbuminemia; AXA 1; AXA1; AXA1; EAOH; EAOH; EOAHA; EOAHA; FHA HIT; FHA HIT; FHA-HIT; FLJ20157; FLJ20157; Forkhead associated domain histidine triad like; Forkhead associated domain histidine triad like; Forkhead associated domain histidine triad like protein; Forkhead-associated domain histidine triad-like protein; MGC1072; MGC1072.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Horse,Rabbit,Sheep,
Applications:	ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800ICC=1:100-500IF=1:100-500（Paraffin sections need antigen repair） not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	41kDa
Cellular localization:	The nucleus
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human Aprataxin:221-320/356
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Storage:	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20°C. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.
PubMed:	PubMed

Product Detail:	Aprataxin is a nuclear protein, present in both the nucleoplasm and the nucleolus, which is a member of the histidine triad (HIT) superfamily. Aprataxin is involved in DNA single-strand break repair, mediating protein-protein interactions with molecules responding to DNA damage. Aprataxin contains three conserved domains: an N-terminal forkhead-associated (FHA) domain which mediates protein-protein interactions, a HIT domain that is similar to Hint, and a C-terminal zinc finger domain. Loss of function mutations in APTX, the gene encoding for Aprataxin, destabilize the Aprataxin protein and result in a rare neurological disorder known as ataxia-oculomotor apraxia, characterized by abnormal movements of the head and eyes. These mutations either target the HIT domain or truncate the protein N-terminal to a zinc finger. **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. **Subunit:** Interacts with single-strand break repair proteins XRCC1, XRCC4, ADPRT and p53/TP53. Interacts with NCL. Interacts (via FHA-like domain) with MDC1 (phosphorylated). **Subcellular Location:** 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. **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. **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.

	Similarity: Contains 1 C2H2-type zinc finger. Contains 1 FHA-like domain. Contains 1 HIT domain. **SWISS:** Q7Z2E3 **Gene ID:** 54840 **Database links:** Entrez Gene: 54840Human Omim: 606350Human SwissProt: Q7Z2E3Human Unigene: 20158Human **Important Note:** This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.