

Aprataxin (B-12): sc-374108

BACKGROUND

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.

REFERENCES

- Gascon, G.G., et al. 1995. Ataxia-oculomotor apraxia syndrome. *J. Child Neurol.* 10: 118-122.
- Online Mendelian Inheritance in Man, OMIM™. 2002. Johns Hopkins University, Baltimore, MD. MIM Number: 606350. World Wide Web URL: <http://www.ncbi.nlm.nih.gov/omim/>
- Gueven, N., et al. 2004. Aprataxin, a novel protein that protects against genotoxic stress. *Hum. Mol. Genet.* 13: 1081-1093.
- Mosesso, P., et al. 2005. The novel human gene Aprataxin is directly involved in DNA single-strand-break repair. *Cell. Mol. Life Sci.* 62: 485-491.
- Crisuolo, C., et al. 2005. Very late onset in ataxia oculomotor apraxia type I. *Ann. Neurol.* 57: 777.
- Ochsner, F., et al. 2005. Mutation of the Aprataxin gene presenting with Charcot-Marie-Tooth-like neuropathy and cerebellar ataxia. *Rev. Neurol.* 161: 331-336.

CHROMOSOMAL LOCATION

Genetic locus: APTX (human) mapping to 9p21.1; Aptx (mouse) mapping to 4 A5.

SOURCE

Aprataxin (B-12) is a mouse monoclonal antibody raised against amino acids 57-356 mapping at the C-terminus of Aprataxin of human origin.

PRODUCT

Each vial contains 200 µg IgG_{2a} kappa light chain in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin. Also available as TransCruz reagent for Gel Supershift and ChIP applications, sc-374108 X, 200 µg/0.1 ml.

Aprataxin (B-12) is available conjugated to agarose (sc-374108 AC), 500 µg/0.25 ml agarose in 1 ml, for IP; to HRP (sc-374108 HRP), 200 µg/ml, for WB, IHC(P) and ELISA; to either phycoerythrin (sc-374108 PE), fluorescein (sc-374108 FITC), Alexa Fluor® 488 (sc-374108 AF488), Alexa Fluor® 546 (sc-374108 AF546), Alexa Fluor® 594 (sc-374108 AF594) or Alexa Fluor® 647 (sc-374108 AF647), 200 µg/ml, for WB (RGB), IF, IHC(P) and FCM; and to either Alexa Fluor® 680 (sc-374108 AF680) or Alexa Fluor® 790 (sc-374108 AF790), 200 µg/ml, for Near-Infrared (NIR) WB, IF and FCM.

APPLICATIONS

Aprataxin (B-12) is recommended for detection of Aprataxin isoforms 1-10 of mouse, rat and human origin by Western Blotting (starting dilution 1:100, dilution range 1:100-1:1000), immunoprecipitation [1-2 µg per 100-500 µg of total protein (1 ml of cell lysate)], immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500), immunohistochemistry (including paraffin-embedded sections) (starting dilution 1:50, dilution range 1:50-1:500) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

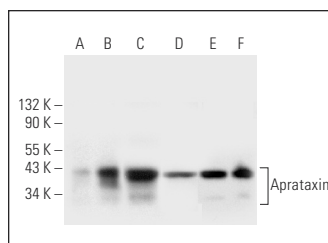
Suitable for use as control antibody for Aprataxin siRNA (h): sc-60196, Aprataxin siRNA (m): sc-60197, Aprataxin shRNA Plasmid (h): sc-60196-SH, Aprataxin shRNA Plasmid (m): sc-60197-SH, Aprataxin shRNA (h) Lentiviral Particles: sc-60196-V and Aprataxin shRNA (m) Lentiviral Particles: sc-60197-V.

Aprataxin (B-12) X TransCruz antibody is recommended for Gel Supershift and ChIP applications.

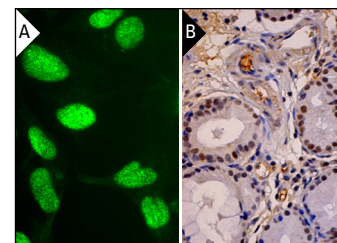
Molecular Weight of Aprataxin isoforms 1-10: 13-41 kDa.

Positive Controls: NCI-H1299 whole cell lysate: sc-364234, HeLa nuclear extract: sc-2120 or Aprataxin (m): 293T Lysate: sc-124980.

DATA



Aprataxin (B-12): sc-374108. Western blot analysis of Aprataxin expression in non-transfected 293T: sc-117752 (A), mouse Aprataxin transfected 293T: sc-124980 (B), NCI-H1299 (C) and Hep G2 (D) whole cell lysates and HeLa (E) and CCRF-CEM (F) nuclear extracts.



Aprataxin (B-12): sc-374108. Immunofluorescence staining of formalin-fixed Hep G2 cells showing nuclear localization (A). Immunoperoxidase staining of formalin fixed, paraffin-embedded human salivary gland tissue showing nuclear staining of glandular cells (B).

SELECT PRODUCT CITATIONS

- Zheng, J., et al. 2019. Diminished OPA1 expression and impaired mitochondrial morphology and homeostasis in Aprataxin-deficient cells. *Nucleic Acids Res.* 47: 4086-4110.
- Ferroni, L., et al. 2019. Effects of novel antidepressant drugs on mesenchymal stem cell physiology. *Biomed. Pharmacother.* 114: 108853.

STORAGE

Store at 4° C, **DO NOT FREEZE**. Stable for one year from the date of shipment. Non-hazardous. No MSDS required.

RESEARCH USE

For research use only, not for use in diagnostic procedures.

Alexa Fluor® is a trademark of Molecular Probes, Inc., Oregon, USA