

Aprataxin (E-9): sc-393648

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

1. Gascon, G.G., et al. 1995. Ataxia-oculomotor apraxia syndrome. *J. Child Neurol.* 10: 118-122.
2. 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/>
3. Gueven, N., et al. 2004. Aprataxin, a novel protein that protects against genotoxic stress. *Hum. Mol. Genet.* 13: 1081-1093.
4. 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.
5. Crisculo, C., et al. 2005. Very late onset in ataxia oculomotor apraxia type I. *Ann. Neurol.* 57: 777.
6. 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 (E-9) is a mouse monoclonal antibody specific for an epitope mapping between amino acids 287-307 near the C-terminus of Aprataxin of human origin.

PRODUCT

Each vial contains 200 µg IgG₁ 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-393648 X, 200 µg/0.1 ml.

Blocking peptide available for competition studies, sc-393648 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% stabilizer protein).

STORAGE

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

APPLICATIONS

Aprataxin (E-9) is recommended for detection of Aprataxin isoforms 1-9 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) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

Aprataxin (E-9) is also recommended for detection of Aprataxin isoforms 1-9 in additional species, including equine, canine, bovine and porcine.

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 (E-9) X TransCruz antibody is recommended for Gel Supershift and ChIP applications.

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

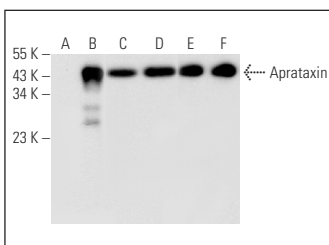
Positive Controls: Aprataxin (m): 293T Lysate: sc-124980, HeLa nuclear extract: sc-2120 or CCRF-CEM nuclear extract: sc-2146.

RECOMMENDED SUPPORT REAGENTS

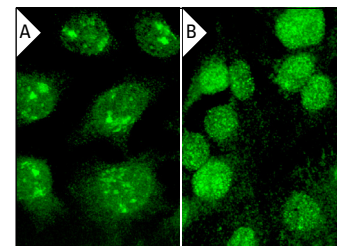
To ensure optimal results, the following support reagents are recommended:

- 1) Western Blotting: use m-IgGκ BP-HRP: sc-516102 or m-IgGκ BP-HRP (Cruz Marker): sc-516102-CM (dilution range: 1:1000-1:10000), Cruz Marker™ Molecular Weight Standards: sc-2035, UltraCruz® Blocking Reagent: sc-516214 and Western Blotting Luminol Reagent: sc-2048.
- 2) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml).
- 3) Immunofluorescence: use m-IgGκ BP-FITC: sc-516140 or m-IgGκ BP-PE: sc-516141 (dilution range: 1:50-1:200) with UltraCruz® Mounting Medium: sc-24941 or UltraCruz® Hard-set Mounting Medium: sc-359850.

DATA



Aprataxin (E-9): sc-393648. 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 (E-9): sc-393648. Immunofluorescence staining of methanol-fixed HeLa (A) and Hep G2 (B) cells showing nucleolar and nuclear localization.

RESEARCH USE

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