

Aprataxin (N-17): sc-48842

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. Criscuolo, 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 (N-17) is an affinity purified goat polyclonal antibody raised against a peptide mapping at the N-terminus of Aprataxin of human origin.

PRODUCT

Each vial contains 200 µg IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-48842 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

Available as TransCruz reagent for Gel Supershift and ChIP applications, sc-48842 X, 200 µg/0.1 ml.

RESEARCH USE

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

APPLICATIONS

Aprataxin (N-17) is recommended for detection of Aprataxin isoforms 1, 3 and 8 of human, mouse and, to a lesser extent, rat origin by Western Blotting (starting dilution 1:200, 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 (N-17) is also recommended for detection of Aprataxin isoforms 1, 3 and 8 in additional species, including equine, 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 (N-17) X TransCruz antibody is recommended for Gel Supershift and ChIP applications.

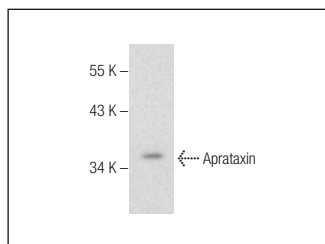
Molecular Weight of Aprataxin isoforms 1-10: 13-41 kDa.

Positive Controls: HeLa nuclear extract: sc-2120 or CCRF-CEM nuclear extract: sc-2146.

RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use donkey anti-goat IgG-HRP: sc-2020 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible donkey anti-goat IgG-HRP: sc-2033 (dilution range: 1:2000-1:5000), Cruz Marker™ Molecular Weight Standards: sc-2035, TBS Blotto A Blocking Reagent: sc-2333 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 donkey anti-goat IgG-FITC: sc-2024 (dilution range: 1:100-1:400) or donkey anti-goat IgG-TR: sc-2783 (dilution range: 1:100-1:400) with UltraCruz™ Mounting Medium: sc-24941.

DATA



Aprataxin (N-17): sc-48842. Western blot analysis of Aprataxin expression in CCRF-CEM nuclear extract.

STORAGE

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