

Aprataxin (m): 293T Lysate: sc-124980

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.
7. Seidle, H.F., et al. 2005. Disease-associated mutations inactivate AMP-lysine hydrolase activity of Aprataxin. *J. Biol. Chem.* 280: 20927-20931.

CHROMOSOMAL LOCATION

Genetic locus: Apx (mouse) mapping to 4 A5.

PRODUCT

Aprataxin (m): 293T Lysate represents a lysate of mouse Aprataxin transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

APPLICATIONS

Aprataxin (m): 293T Lysate is suitable as a Western Blotting positive control for mouse reactive Aprataxin antibodies. Recommended use: 10-20 µl per lane.

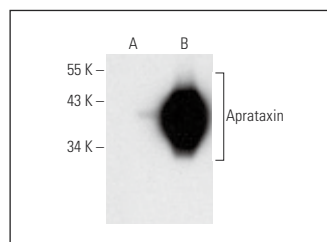
Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

Aprataxin (B-3): sc-365849 is recommended as a positive control antibody for Western Blot analysis of enhanced mouse Aprataxin expression in Aprataxin transfected 293T cells (starting dilution 1:100, dilution range 1:100-1:1,000).

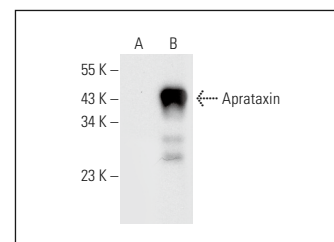
RECOMMENDED SUPPORT REAGENTS

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.

DATA



Aprataxin (B-3): sc-365849. Western blot analysis of Aprataxin expression in non-transfected: sc-117752 (A) and mouse Aprataxin transfected: sc-124980 (B) 293T whole cell lysates.



Aprataxin (E-9): sc-393648. Western blot analysis of Aprataxin expression in non-transfected: sc-117752 (A) and mouse Aprataxin transfected: sc-124980 (B) 293T whole cell lysates.

STORAGE

Store at -20° C. Repeated freezing and thawing should be minimized. Sample vial should be boiled once prior to use. Non-hazardous. No MSDS required.

RESEARCH USE

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

PROTOCOLS

See our web site at www.scbt.com for detailed protocols and support products.