SANTA CRUZ BIOTECHNOLOGY, INC.

SMCHD1 (A-16): sc-139554

BACKGROUND

SMCHD1 (structural maintenance of chromosomes flexible hinge domain containing 1) is a 2,005 amino acid protein and novel modifier of epigenetic gene silencing. SMCHD1 may have a role in the hypermethylation of CpG islands associated with the maintenance of X chromosome inactivation. SMCHD1 exists as three alternatively spliced isoforms and is encoded by a gene located on human chromosome 18, which houses over 300 protein-coding genes and contains nearly 76 million bases, representing about 2.5% of total DNA in cells. There are a variety of diseases associated with defects in chromosome 18-localized genes, some of which include Niemann-Pick disease, hereditary hemorrhagic telangiectasia, erythropoietic protoporphyria and follicular lymphomas. Also, three chromosomal abnormalities result from meiotic nondisjunction events of chromosome 18: Monosomy 18p, Trisomy 18 (also known as Edwards syndrome) and Tetrasomy 18p.

REFERENCES

1. Grosso, S., et al. 2005. Chromosome 18 aberrations and epilepsy: a review. Am. J. Med. Genet. A 134A: 88-94.
2. Cerminara, C., et al. 2008. Epilepsy and deletion syndromes of chromosome 18: do not forget the short arm! Epilepsia 49: 1813-1814.
3. Ashe, A., et al. 2008. A genome-wide screen for modifiers of transgene variegation identifies genes with critical roles in development. Genome Biol. 9: R182.
4. Kohan, R., et al. 2008. Improving the health care experiences of families given the prenatal diagnosis of Trisomy 18. J. Perinatol. 28: 719.
5. Blewitt, M.E., et al. 2008. SmcHD1, containing a structural-maintenance-of-chromosomes hinge domain, has a critical role in X inactivation. Nat. Genet. 40: 663-669.
6. Shaw, J. 2008. Trisomy 18: a case study. Neonatal Netw. 27: 33-41.
7. Turleau, C. 2008. Monosomy 18p. Orphanet J. Rare Dis. 3: 4.
8. Witters, I., et al. 2008. Trisomy 18 presenting with severe limb deformations. Prenat. Diagn. 28: 549-550.
9. Edwards, S., et al. 2008. Prenatal diagnosis of monosomy 18p involving a jumping translocation. Prenat. Diagn. 28: 764-766.

CHROMOSOMAL LOCATION

Genetic locus: SMCHD1 (human) mapping to 18p11.32; Smchd1 (mouse) mapping to 17 E1.3.

SOURCE

SMCHD1 (A-16) is an affinity purified rabbit polyclonal antibody raised against a peptide mapping within an internal region of SMCHD1 of human origin.

PRODUCT

Each vial contains 100 µg IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-139554 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

SMCHD1 (A-16) is recommended for detection of SMCHD1 of mouse, rat and human origin by Western Blotting (starting dilution 1:100, dilution range 1:50-1:500), immunoprecipitation [1-2 µg per 100-500 µg of total protein (1 ml of cell lysate)], immunofluorescence (starting dilution 1:25, dilution range 1:25-1:250) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

SMCHD1 (A-16) is also recommended for detection of SMCHD1 in additional species, including equine, canine, bovine and avian.

Suitable for use as control antibody for SMCHD1 siRNA (h): sc-156012, Smchd1 siRNA (m): sc-153621, SMCHD1 shRNA Plasmid (h): sc-156012-SH, Smchd1 shRNA Plasmid (m): sc-153621-SH, SMCHD1 shRNA (h) Lentiviral Particles: sc-156012-V and Smchd1 shRNA (m) Lentiviral Particles: sc-153621-V.

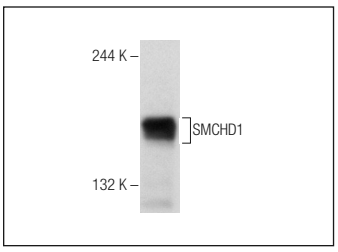
Molecular Weight of SMCHD1: 226 kDa.

Positive Controls: mouse brain extract: sc-2253.

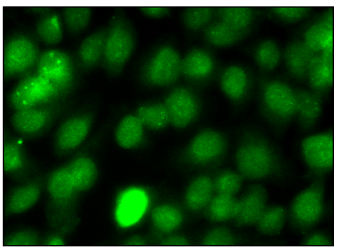
RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use goat anti-rabbit IgG-HRP: sc-2004 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible goat anti-rabbit IgG-HRP: sc-2030 (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 goat anti-rabbit IgG-FITC: sc-2012 (dilution range: 1:100-1:400) or goat anti-rabbit IgG-TR: sc-2780 (dilution range: 1:100-1:400) with UltraCruz™ Mounting Medium: sc-24941.

DATA



SMCHD1 (A-16): sc-139554. Western blot analysis of SMCHD1 expression in mouse brain tissue extract.



SMCHD1 (A-16): sc-139554. Immunofluorescence staining of formalin-fixed HeLa cells showing nuclear localization. Kindly provided by Yang Xiang, Ph.D., Division of Newborn Medicine, Boston Children's Hospital, Cell Biology Department, Harvard Medical School.

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.