

# HYLS1 (h): 293 Lysate: sc-111891

## BACKGROUND

The hydrolethalus syndrome protein 1 (HYLS1) is a widely conserved protein that plays an essential role in cilia formation. A single amino acid mutation in the HYLS1 gene leads to a perinatal lethal disorder termed hydrolethalus syndrome, a severe fetal malformation syndrome characterized by central nervous system (CNS) malformation such as hydrocephaly and absent midline structures of the brain, micrognathia, defective lobation of the lungs and polydactyly. The gene encoding HYLS1 maps to human chromosome 11, which makes up around 4% of human genomic DNA and is considered a gene and disease association dense chromosome. The chromosome 11 encoded Atm gene is important for regulation of cell cycle arrest and apoptosis following double strand DNA breaks. Atm mutation leads to the disorder known as ataxia-telangiectasia.

## REFERENCES

1. Grossfeld, P.D., et al. 2004. The 11q terminal deletion disorder: a prospective study of 110 cases. *Am. J. Med. Genet. A* 129A: 51-61.
2. Mee, L., et al. 2005. Hydrolethalus syndrome is caused by a missense mutation in a novel gene HYLS1. *Hum. Mol. Genet.* 14: 1475-1488.
3. Taylor, T.D., et al. 2006. Human chromosome 11 DNA sequence and analysis including novel gene identification. *Nature* 440: 497-500.
4. Lee, S.P., et al. 2007. Phase I study of eptifibatide in patients with sickle cell anaemia. *Br. J. Haematol.* 139: 612-620.
5. Lee, J.H. and Paull, T.T. 2007. Activation and regulation of Atm kinase activity in response to DNA double-strand breaks. *Oncogene* 26: 7741-7748.
6. Paetau, A., et al. 2008. Hydrolethalus syndrome: neuropathology of 21 cases confirmed by HYLS1 gene mutation analysis. *J. Neuropathol. Exp. Neurol.* 67: 750-762.
7. Kaste, S.C., et al. 2008. Wilms tumour: prognostic factors, staging, therapy and late effects. *Pediatr. Radiol.* 38: 2-17.
8. Dammermann, A., et al. 2009. The hydrolethalus syndrome protein HYLS1 links core centriole structure to cilia formation. *Genes Dev.* 23: 2046-2059.
9. Honkala, H., et al. 2009. Unraveling the disease pathogenesis behind lethal hydrolethalus syndrome revealed multiple changes in molecular and cellular level. *Pathogenetics* 2: 2.

## CHROMOSOMAL LOCATION

Genetic locus: HYLS1 (human) mapping to 11q24.2.

## PRODUCT

HYLS1 (h): 293 Lysate represents a lysate of human HYLS1 transfected 293 cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

## 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.

## APPLICATIONS

HYLS1 (h): 293 Lysate is suitable as a Western Blotting positive control for human reactive HYLS1 antibodies. Recommended use: 10-20 µl per lane.

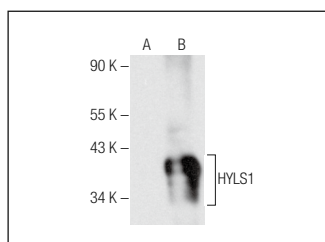
Control 293 Lysate: sc-110760 is available as a Western Blotting negative control lysate derived from non-transfected 293 cells.

HYLS1 (D-9): sc-376721 is recommended as a positive control antibody for Western Blot analysis of enhanced human HYLS1 expression in HYLS1 transfected 293 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



HYLS1 (D-9): sc-376721. Western blot analysis of HYLS1 expression in non-transfected: sc-110760 (A) and human HYLS1 transfected: sc-111891 (B) 293 whole cell lysates.

## RESEARCH USE

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

## PROTOCOLS

See our web site at [www.scbt.com](http://www.scbt.com) for detailed protocols and support products.