

HYLS1 (E-3): sc-398834

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

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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.
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CHROMOSOMAL LOCATION

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

SOURCE

HYLS1 (E-3) is a mouse monoclonal antibody raised against amino acids 1-299 representing full length HYLS1 of human origin.

PRODUCT

Each vial contains 200 µg IgG_{2b} kappa light chain in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

APPLICATIONS

HYLS1 (E-3) is recommended for detection of HYLS1 of 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).

Suitable for use as control antibody for HYLS1 siRNA (h): sc-96710, HYLS1 shRNA Plasmid (h): sc-96710-SH and HYLS1 shRNA (h) Lentiviral Particles: sc-96710-V.

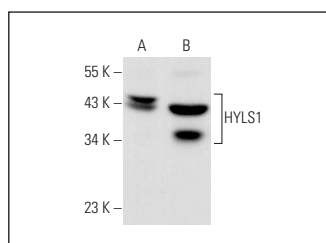
Molecular Weight of HYLS1: 40 kDa.

Positive Controls: HYLS1 (h): 293 Lysate: sc-111891, PC-3 cell lysate: sc-2220 or THP-1 cell lysate: sc-2238.

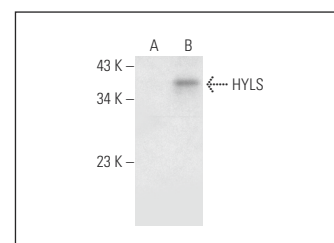
RECOMMENDED SUPPORT REAGENTS

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



HYLS1 (E-3): sc-398834. Western blot analysis of HYLS1 expression in PC-3 (A) and THP-1 (B) whole cell lysates.



HYLS1 (E-3): sc-398834. Western blot analysis of HYLS1 expression in non-transfected: sc-110760 (A) and human HYLS1 transfected: sc-111891 (B) 293 whole cell lysates.

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.

PROTOCOLS

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