SANTA CRUZ BIOTECHNOLOGY, INC.

NSDHL (B-5): sc-390994

SANTA CRUZ BIOTECHNOLOGY

The Power to Question

BACKGROUND

NSDHL (NAD(P) dependent steroid dehydrogenase-like), also known as H105E3, XAP104 or SDR31E1, is a 373 amino acid single-pass membrane protein expressed in brain, heart, liver, lung, kidney, skin and placenta. Belonging to the 3-β-HSD family, NSDHL localizes to the endoplasmic reticulum and is involved in cholesterol biosynthesis. NSDHL is thought to be involved in the demethylation of sterol precursors in one of the later steps of cholesterol biosynthesis. Defects in the gene encoding NSDHL causes congenital hemidysplasia with ichthyosiform erythroderma and limb defects (CHILD), which is an X-linked dominant disorder of lipid metabolism with defective cholesterol biosynthesis that usually results in male lethality. CHILD is characterized by congenital hemidysplasia with ichthyosiform erythroderma and ipsilateral hypoplasia of limbs and other parts of the skeleton.

REFERENCES

1. Hummel, M., et al. 2003. Left-sided CHILD syndrome caused by a nonsense mutation in the NSDHL gene. Am. J. Med. Genet. A 122A: 246-251.
2. Caldas, H., et al. 2003. NSDHL, an enzyme involved in cholesterol biosynthesis, traffics through the Golgi and accumulates on ER membranes and on the surface of lipid droplets. Hum. Mol. Genet. 12: 2981-2991.
3. Ohashi, M., et al. 2003. Localization of mammalian NAD(P)H steroid dehydrogenase-like protein on lipid droplets. J. Biol. Chem. 278: 36819-36829.
4. Murata, K., et al. 2003. A unique point mutation in the NSDHL gene in a Japanese patient with CHILD syndrome. J. Dermatol. Sci. 33: 67-69.
5. Mehra, S., et al. 2005. A novel somatic mutation of the 3β-hydroxysteroid dehydrogenase gene in sporadic cutaneous verruciform xanthoma. Arch. Dermatol. 141: 1263-1267.
6. Bittar, M., et al. 2006. CHILD syndrome in 3 generations: the importance of mild or minimal skin lesions. Arch. Dermatol. 142: 348-351.
7. Guggenberger, C., et al. 2007. Functional analysis of cholesterol biosynthesis by RNA interference. J. Steroid Biochem. Mol. Biol. 104: 105-109.
8. Saito, M., et al. 2008. A novel silent mutation in the NSDHL gene causing CHILD syndrome as a result of aberrant splicing. Br. J. Dermatol. 159: 1204-1206.
9. Cunningham, D., et al. 2009. Developmental expression pattern of the cholesterogenic enzyme NSDHL and negative selection of NSDHL-deficient cells in the heterozygous Bpa^{1H}/+ mouse. Mol. Genet. Metab. 98: 356-366.

CHROMOSOMAL LOCATION

Genetic locus: NSDHL (human) mapping to Xq28.

SOURCE

NSDHL (B-5) is a mouse monoclonal antibody raised against amino acids 1-195 mapping at the N-terminus of NSDHL of human origin.

RESEARCH USE

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

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

NSDHL (B-5) is recommended for detection of NSDHL 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), immunohistochemistry (including paraffin-embedded sections) (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 NSDHL siRNA (h): sc-90849, NSDHL shRNA Plasmid (h): sc-90849-SH and NSDHL shRNA (h) Lentiviral Particles: sc-90849-V.

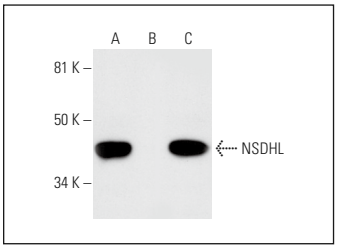
Molecular Weight of NSDHL: 42 kDa.

Positive Controls: A-431 whole cell lysate: sc-2201 or HeLa whole cell lysate: sc-2200.

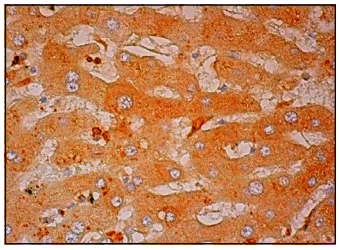
RECOMMENDED SUPPORT REAGENTS

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. 4) Immunohistochemistry: use m-IgGκ BP-HRP: sc-516102 with DAB, 50X: sc-24982 and Immunohistomount: sc-45086, or Organo/Limonene Mount: sc-45087.

DATA



NSDHL (B-5): sc-390994. Western blot analysis of NSDHL expression in A-431 (**A**), F9 (**B**) and HeLa (**C**) whole cell lysates. Note lack of reactivity in lane **B**.



NSDHL (B-5): sc-390994. Immunoperoxidase staining of formalin fixed, paraffin-embedded human liver tissue showing cytoplasmic staining of hepatocytes.

STORAGE

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