

NSDHL (m): 293T Lysate: sc-122135

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

- Hummel, M., Cunningham, D., Mullett, C.J., Kelley, R.I. and Herman, G.E. 2003. Left-sided CHILD syndrome caused by a nonsense mutation in the NSDHL gene. *Am. J. Med. Genet. A* 122A: 246-251.
- Caldas, H. and Herman, G.E. 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.
- Ohashi, M., Mizushima, N., Kabeya, Y. and Yoshimori, T. 2003. Localization of mammalian NAD(P)H steroid dehydrogenase-like protein on lipid droplets. *J. Biol. Chem.* 278: 36819-36829.
- Murata, K., Shinkai, H., Ishikiriya, S., Yamazaki, M., Fukuzumi, Y. and Hatamochi, A. 2003. A unique point mutation in the NSDHL gene in a Japanese patient with CHILD syndrome. *J. Dermatol. Sci.* 33: 67-69.
- Mehra, S., Li, L., Fan, C.Y., Smoller, B., Morgan, M. and Somach, S. 2005. A novel somatic mutation of the 3 β -hydroxysteroid dehydrogenase gene in sporadic cutaneous verruciform xanthoma. *Arch. Dermatol.* 141: 1263-1267.
- Bittar, M., Happle, R., Grzeschik, K.H., Leveleki, L., Hertl, M., Bornholdt, D. and Konig, A. 2006. CHILD syndrome in 3 generations: the importance of mild or minimal skin lesions. *Arch. Dermatol.* 142: 348-351.
- Guggenberger, C., Ilgen, D. and Adamski, J. 2007. Functional analysis of cholesterol biosynthesis by RNA interference. *J. Steroid Biochem. Mol. Biol.* 104: 105-109.
- Saito, M. and Ishiko, A. 2008. A novel silent mutation in the NSDHL gene causing CHILD syndrome as a result of aberrant splicing. *Br. J. Dermatol.* 159: 1204-1206.
- Cunningham, D., Sychala, K., McLarren, K.W., Garza, L.A., Boerkoel, C.F. and Herman, G.E. 2009. Developmental expression pattern of the cholesterologenic 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 (mouse) mapping to X A7.3.

PRODUCT

NSDHL (m): 293T Lysate represents a lysate of mouse NSDHL transfected 293T cells and is provided as 100 μ g protein in 200 μ l SDS-PAGE buffer.

APPLICATIONS

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

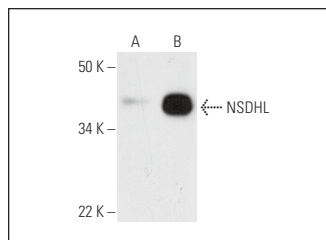
Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

NSDHL (D-11): sc-390871 is recommended as a positive control antibody for Western Blot analysis of enhanced mouse NSDHL expression in NSDHL 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-IgG κ BP-HRP: sc-516102 or m-IgG κ BP-HRP (Cruz Marker): sc-516102-CM (dilution range: 1:1000-1:10000), Cruz MarkerTM Molecular Weight Standards: sc-2035, UltraCruz[®] Blocking Reagent: sc-516214 and Western Blotting Luminol Reagent: sc-2048.

DATA



NSDHL (D-11): sc-390871. Western blot analysis of NSDHL expression in non-transfected: sc-117752 (A) and mouse NSDHL transfected: sc-122135 (B) 293T whole cell lysates.

STORAGE

Store at -20 $^{\circ}$ 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.