

Sc5d (C-14): sc-55893

BACKGROUND

Sc5d (sterol-C5-desaturase), also known as SC5DL, ERG3 or S5DES, is a 299 amino acid multi-pass membrane protein that localizes to the endoplasmic reticulum and belongs to the sterol desaturase family. Using iron as a cofactor, Sc5d functions to catalyze a dehydrogenation reaction that introduces a C5-6 double bond into lathosterol, a process that is NADPH- and oxygen-dependent. Defects in the gene encoding Sc5d are the cause of lathosterolosis, an autosomal recessive disorder that is characterized by liver disease, mental retardation and congenital anomalies. The gene encoding Sc5d maps to human chromosome 11, which houses over 1,400 genes and comprises nearly 4% of the human genome. Jervell and Lange-Nielsen syndrome, Jacobsen syndrome, Niemann-Pick disease, hereditary angioedema and Smith-Lemli-Opitz syndrome are associated with defects in genes that maps to chromosome 11.

REFERENCES

1. Ves-Losada, A., et al. 1995. Fatty acid δ 5 desaturation in rat liver cell nuclei. *Mol. Cell. Biochem.* 142: 163-170.
2. Matsushima, M., et al. 1996. Molecular cloning and mapping of a human cDNA (SC5DL) encoding a protein homologous to fungal sterol-C5-desaturase. *Cytogenet. Cell Genet.* 74: 252-254.
3. Cho, H.P., et al. 1999. Cloning, expression, and fatty acid regulation of the human δ -5 desaturase. *J. Biol. Chem.* 274: 37335-37339.
4. Nishi, S., et al. 2000. cDNA cloning of the mammalian sterol C5-desaturase and the expression in yeast mutant. *Biochim. Biophys. Acta* 1490: 106-108.
5. Sugawara, T., et al. 2001. Molecular cloning and structural analysis of human sterol C5 desaturase. *Biochim. Biophys. Acta* 1533: 277-284.
6. Brunetti-Pierri, N., et al. 2002. Lathosterolosis, a novel multiple-malformation/mental retardation syndrome due to deficiency of 3 β -hydroxysteroid-delta5-desaturase. *Am. J. Hum. Genet.* 71: 952-958.
7. Krakowiak, P.A., et al. 2003. Lathosterolosis: an inborn error of human and murine cholesterol synthesis due to lathosterol 5-desaturase deficiency. *Hum. Mol. Genet.* 12: 1631-1641.
8. Lu, Y., et al. 2008. Multiple genetic variants along candidate pathways influence plasma high-density lipoprotein cholesterol concentrations. *J. Lipid Res.* 49: 2582-2589.
9. Online Mendelian Inheritance in Man, OMIM™. 2008. Johns Hopkins University, Baltimore, MD. MIM Number: 602286. World Wide Web URL: <http://www.ncbi.nlm.nih.gov/omim/>

CHROMOSOMAL LOCATION

Genetic locus: SC5DL (human) mapping to 11q23.3.

SOURCE

Sc5d (C-14) is an affinity purified goat polyclonal antibody raised against a peptide mapping at the C-terminus of Sc5d of human origin.

PRODUCT

Each vial contains 200 μ g IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-55893 P, (100 μ g peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

Sc5d (C-14) is recommended for detection of Sc5d of human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunoprecipitation [1-2 μ g per 100-500 μ g 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 Sc5d siRNA (h): sc-62980, Sc5d shRNA Plasmid (h): sc-62980-SH and Sc5d shRNA (h) Lentiviral Particles: sc-62980-V.

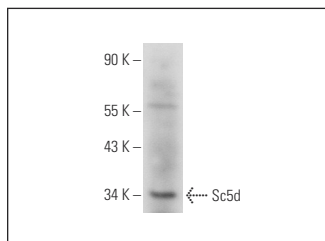
Molecular Weight of Sc5d: 35 kDa.

Positive Controls: human liver extract: sc-363766.

RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use donkey anti-goat IgG-HRP: sc-2020 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible donkey anti-goat IgG-HRP: sc-2033 (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 donkey anti-goat IgG-FITC: sc-2024 (dilution range: 1:100-1:400) or donkey anti-goat IgG-TR: sc-2783 (dilution range: 1:100-1:400) with UltraCruz™ Mounting Medium: sc-24941.

DATA



Sc5d (C-14): sc-55893. Western blot analysis of Sc5d expression in human liver tissue extract.

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.