ABHD5 (h): 293 Lysate: sc-112234



The Power to Question

BACKGROUND

 α/β -hydrolase domains are characterized by a catalytic triad composed of a histidine, an acid and a nucleophile. ABHD5 (α/β -hydrolase domain-containing 5), also known as CGI-58, NCIE2 or CDS, is a 349 amino acid protein that contains an α/β -hydrolase domain, through which it conveys catalytic function. Localized to the surface of lipid droplets, ABHD5 is widely expressed and interacts with Perilipin on the surface of lipid droplets where it facilitates lipolysis, the breakdown of fat. Defects in the gene encoding ABHD5 are the cause of Chanarin-Dorfman syndrome (CDS), an autosomal recessive inbornerror of lipid metabolism with impaired long-chain fatty acid oxidation. CDS symptoms include congenital generalized ichthyosis, vacuolated leukocytes, hepatomegaly, myopathy, cataracts, neurosensory hearing loss and developmental delay.

REFERENCES

- Lefèvre, C., Jobard, F., Caux, F., Bouadjar, B., Karaduman, A., Heilig, R., Lakhdar, H., Wollenberg, A., Verret, J.L., Weissenbach, J., Ozgüc, M., Lathrop, M., Prud'homme, J.F. and Fischer, J. 2001. Mutations in CGI-58, the gene encoding a new protein of the esterase/lipase/thioesterase subfamily, in Chanarin-Dorfman syndrome. Am. J. Hum. Genet. 69: 1002-1012.
- Schleinitz, N., Fischer, J., Sanchez, A., Veit, V., Harle, J.R. and Pelissier, J.F. 2005. Two new mutations of the ABHD5 gene in a new adult case of Chanarin-Dorfman syndrome: an uncommon lipid storage disease. Arch. Dermatol. 141: 798-800.
- Lass, A., Zimmermann, R., Haemmerle, G., Riederer, M., Schoiswohl, G., Schweiger, M., Kienesberger, P., Strauss, J.G., Gorkiewicz, G. and Zechner, R. 2006. Adipose triglyceride lipase-mediated lipolysis of cellular fat stores is activated by CGI-58 and defective in Chanarin-Dorfman syndrome. Cell Metab. 3: 309-319.
- Ben Selma, Z., Yilmaz, S., Schischmanoff, P.O., Blom, A., Ozogul, C., Laroche, L. and Caux, F. 2007. A novel S115G mutation of CGI-58 in a Turkish patient with Dorfman-Chanarin syndrome. J. Invest. Dermatol. 127: 2273-2276.
- 5. Fischer, J., Negre-Salvayre, A. and Salvayre, R. 2007. Neutral lipid storage diseases and ATGL (adipose triglyceride lipase) and CGI-58/ABHD5 (α/β hydrolase domain-containing 5) deficiency: myopathy, ichthyosis, but no obesity. Med. Sci. 23: 575-578.
- Yamaguchi, T., Omatsu, N., Morimoto, E., Nakashima, H., Ueno, K., Tanaka, T., Satouchi, K., Hirose, F. and Osumi, T. 2007. CGI-58 facilitates lipolysis on lipid droplets but is not involved in the vesiculation of lipid droplets caused by hormonal stimulation. J. Lipid Res. 48: 1078-1089.
- 7. Online Mendelian Inheritance in Man, OMIM™. 2007. Johns Hopkins University, Baltimore, MD. MIM Number: 604780. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/

CHROMOSOMAL LOCATION

Genetic locus: ABHD5 (human) mapping to 3p21.33.

PRODUCT

ABHD5 (h): 293 Lysate represents a lysate of human ABHD5 transfected 293 cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

APPLICATIONS

ABHD5 (h): 293 Lysate is suitable as a Western Blotting positive control for human reactive ABHD5 antibodies. Recommended use: 10-20 µl per lane.

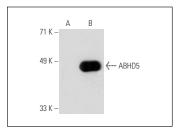
Control 293 Lysate: sc-110760 is available as a Western Blotting negative control lysate derived from non-transfected 293 cells.

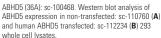
ABHD5 (36A): sc-100468 is recommended as a positive control antibody for Western Blot analysis of enhanced human ABHD5 expression in ABHD5 transfected 293 cells (starting dilution 1:100, dilution range 1:100-1:1,000).

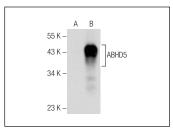
RECOMMENDED SUPPORT REAGENTS

To ensure optimal results, the following support reagents are recommended: 1) Western Blotting: use m-lgG κ BP-HRP: sc-516102 or m-lgG 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







ABHD5 (C-9): sc-377270. Western blot analysis of ABHD5 expression in non-transfected: sc-110760 (A) and human ABHD5 transfected: sc-112234 (B) 293 whole cell Ivsates.

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.

Santa Cruz Biotechnology, Inc. 1.800.457.3801 831.457.3801 **Europe** +00800 4573 8000 49 6221 4503 0 **www.scbt.com**