SANTA CRUZ BIOTECHNOLOGY, INC.

SHFM3 (C-17): sc-130266



The Power to Question

BACKGROUND

SHFM3 (split-hand/foot malformation type 3), also known as FBXW4 (F-box and WD-40 domain-containing protein 4) or dactylin, is a 412 amino acid protein that is primarily involved in signaling pathways that are essential for normal limb development. Specifically, SHFM3 promotes ubiquination and degradation of certain phosphorylated proteins. Defects in the gene encoding SHFM3 are the cause of split-hand/foot malformation type 3, an autosomal dominant disorder that is characterized by hypoplasia/aplasia of the central digits, causing variable fusion with the remaining digits. The disease is not a result of a point mutation, but rather a genomic rearrangement resulting in a tandem duplication that contains a disrupted copy of the gene encoding SHFM3, as well as extra copies of two other genes that are linked to limb development. SHFM3 is typically expressed in kidney, brain, lung and liver, with highest expression in fetal brain.

REFERENCES

- Ianakiev, P., Kilpatrick, M.W., Dealy, C., Kosher, R., Korenberg, J.R., Chen, X.N. and Tsipouras, P. 1999. A novel human gene encoding an F-box/WD40 containing protein maps in the SHFM3 critical region on 10q24. Biochem. Biophys. Res. Commun. 261: 64-70.
- Sidow, A., Bulotsky, M.S., Kerrebrock, A.W., Birren, B.W., Altshuler, D., Jaenisch, R., Johnson, K.R. and Lander, E.S. 1999. A novel member of the F-box/WD40 gene family, encoding dactylin, is disrupted in the mouse dactylaplasia mutant. Nat. Genet. 23: 104-107.
- 3. Online Mendelian Inheritance in Man, OMIM™. 2002. Johns Hopkins University, Baltimore, MD. MIM Number: 608071. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/
- Basel, D., DePaepe, A., Kilpatrick, M.W. and Tsipouras, P. 2003. Split hand foot malformation is associated with a reduced level of dactylin gene expression. Clin. Genet. 64: 350-354.
- de Mollerat, X.J., Gurrieri, F., Morgan, C.T., Sangiorgi, E., Everman, D.B., Gaspari, P., Amiel, J., Bamshad, M.J., Lyle, R., Blouin, J.L., Allanson, J.E., Le Marec, B., Wilson, M., Braverman, N.E., Radhakrishna, U., Delozier-Blanchet, C., Abbott, A., Elghouzzi, V., Antonarakis, S., Stevenson, R.E., Munnich, A., Neri, G. and Schwartz, C.E. 2003. A genomic rearrangement resulting in a tandem duplication is associated with split hand-split foot malformation 3 (SHFM3) at 10q24. Hum. Mol. Genet. 12: 1959-1971.
- Roscioli, T., Taylor, P.J., Bohlken, A., Donald, J.A., Masel, J., Glass, I.A. and Buckley, M.F. 2004. The 10q24-linked split hand/split foot syndrome (SHFM3): narrowing of the critical region and confirmation of the clinical phenotype. Am. J. Med. Genet. A. 124A: 136-141.
- Kano, H., Kurosawa, K., Horii, E., Ikegawa, S., Yoshikawa, H., Kurahashi, H. and Toda, T. 2005. Genomic rearrangement at 10q24 in non-syndromic split-hand/split-foot malformation. Hum. Genet. 118: 477-483.

CHROMOSOMAL LOCATION

Genetic locus: FBXW4 (human) mapping to 10q24.32.

RESEARCH USE

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

SOURCE

SHFM3 (C-17) is a purified rabbit polyclonal antibody raised against a peptide mapping near the C-terminus of SHFM3 of human origin.

PRODUCT

Each vial contains 100 μg lgG in 1.0 ml PBS with < 0.1% sodium azide and 0.1% gelatin.

APPLICATIONS

SHFM3 (C-17) is recommended for detection of SHFM3 of human origin by 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).

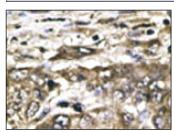
Suitable for use as control antibody for SHFM3 siRNA (h): sc-90449, SHFM3 shRNA Plasmid (h): sc-90449-SH and SHFM3 shRNA (h) Lentiviral Particles: sc-90449-V.

Molecular Weight of SHFM3: 46 kDa.

RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Immunofluorescence: use goat anti-rabbit IgG-FITC: sc-2012 (dilution range: 1:100-1:400) or goat anti-rabbit IgG-TR: sc-2780 (dilution range: 1:100-1:400) with UltraCruz™ Mounting Medium: sc-24941. 2) Immunohistochemistry: use ImmunoCruz™: sc-2051 or ABC: sc-2018 rabbit IgG Staining Systems.

DATA



SHFM3 (C-17): sc-130266. Immunoperoxidase staining of formalin fixed, paraffin-embedded human cancer tissue showing cytoplasmic staining.

STORAGE

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

PROTOCOLS

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