

EVC (H-300): sc-134873

BACKGROUND

EVC, or Ellis-van Creveld syndrome, is an autosomal skeletal dysplasia caused by mutations in the EVC and EVC2 genes. Found in developing ribs, heart, kidney and lung, the EVC gene is responsible for normal development of the face, limbs, teeth and nails. The protein expressed by the EVC gene is an intracellular component of the hedgehog signal pathway that contains a leucine zipper and transmembrane domain. Defects in the EVC gene can lead to short-limb dwarfism, ectodermal dysplasia and cardiac anomalies such as irregular atrioventricular septum development. Additionally, the EVC gene has been implicated in Weyers acrodistal dysostosis, an autosomal dominant disease characterized by facial abnormalities and limb defects.

REFERENCES

1. Polymeropoulos, M.H., Ide, S.E., Wright, M., Goodship, J., Weissenbach, J., Pyeritz, R.E., Da Silva, E.O., Ortiz De Luna, R.I. and Francomano, C.A. 1996. The gene for the Ellis-van Creveld syndrome is located on chromosome 4p16. *Genomics* 35: 1-5.
2. Ruiz-Perez, V.L., Ide, S.E., Strom, T.M., Lorenz, B., Wilson, D., Woods, K., King, L., Francomano, C., Freisinger, P., Spranger, S., Marino, B., Dallapiccola, B., Wright, M., Meitinger, T., Polymeropoulos, M.H. and Goodship, J. 2000. Mutations in a new gene in Ellis-van Creveld syndrome and Weyers acrodistal dysostosis. *Nat. Genet.* 24: 283-286.
3. Galdzicka, M., Patnala, S., Hirshman, M.G., Cai, J.F., Nitowsky, H., Egeland, J.A. and Ginns, E.I. 2002. A new gene, EVC is mutated in Ellis-van Creveld syndrome. *Mol. Genet. Metab.* 77: 291-295.
4. Mostafa, M.I., Temtamy, S.A., el-Gammal, M.A. and Mazen, I.M. 2005. Unusual pattern of inheritance and orofacial changes in the Ellis-van Creveld syndrome. *Genet. Couns.* 16: 75-83.
5. van Hagen, J.M., Baart, J.A. and Gille, J.J. 2005. From gene to disease; EVC, EVC2, and Ellis-van Creveld syndrome. *Ned. Tijdschr. Geneesk.* 149: 929-931.
6. Ye, X., Song, G., Fan, M., Shi, L., Jabs, E.W., Huang, S., Guo, R. and Bian, Z. 2006. A novel heterozygous deletion in the EVC2 gene causes Weyers acrofacial dysostosis. *Hum. Genet.* 119: 199-205.

CHROMOSOMAL LOCATION

Genetic locus: EVC (human) mapping to 4p16.2; Evc (mouse) mapping to 5 B3.

SOURCE

EVC (H-300) is a rabbit polyclonal antibody raised against amino acids 181-480 mapping within an internal region of EVC of human origin.

PRODUCT

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

STORAGE

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

APPLICATIONS

EVC (H-300) is recommended for detection of EVC of human and, to a lesser extent, mouse and rat origin by Western Blotting (starting dilution 1:200, 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) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

Suitable for use as control antibody for EVC siRNA (h): sc-72235, EVC siRNA (m): sc-72236, EVC shRNA Plasmid (h): sc-72235-SH, EVC shRNA Plasmid (m): sc-72236-SH, EVC shRNA (h) Lentiviral Particles: sc-72235-V and EVC shRNA (m) Lentiviral Particles: sc-72236-V.

Molecular Weight of EVC: 124 kDa.

Positive Controls: MCF7 whole cell lysate: sc-2206.

RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use goat anti-rabbit IgG-HRP: sc-2004 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible goat anti-rabbit IgG-HRP: sc-2030 (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 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.

RESEARCH USE

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

PROTOCOLS

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