

EVC2 (F-12): sc-393128



The Power to Question

BACKGROUND

Ellis van Creveld syndrome 2 (EVC2), also designated limb1n, is a protein containing a leucine zipper and a transmembrane domain. EVC2 is expressed in the developing vertebral bodies, kidney, ribs, lung, upper and lower limbs and heart. This protein is implicated in two major diseases: Ellis-van Creveld syndrome (EVC) and Weyers acrodistal dysostosis (WAD). EVC is characterized by short-limb dwarfism, short ribs and dysplastic nails and teeth. It is an autosomal recessive disorder often causing heart defects. WAD is an autosomal dominant disorder and although the phenotype of WAD is milder than EVC, it still causes dysplastic nails, short limbs and short stature.

REFERENCES

1. Brueton, L.A., et al. 1990. Ellis-van Creveld syndrome, Jeune syndrome, and renal-hepatic-pancreatic dysplasia: separate entities or disease spectrum? *J. Med. Genet.* 27: 252-255.
2. Ide, S.E., et al. 1996. Exclusion of the MSX1 homeobox gene as the gene for the Ellis van Creveld syndrome in the Amish. *Hum. Genet.* 98: 572-575.
3. McKusick, V.A. 2000. Ellis-van Creveld syndrome and the Amish. *Nat. Genet.* 24: 203-204.
4. Arya, L., et al. 2001. Ellis-van Creveld syndrome: a report of two cases. *Pediatr. Dermatol.* 18: 485-489.
5. Tompson, S.W., et al. 2001. Ellis-van Creveld syndrome resulting from segmental uniparental disomy of chromosome 4. *J. Med. Genet.* 38: E18.
6. Sajeev, C.G., et al. 2002. Images in cardiology: common atrium in a child with Ellis-van Creveld syndrome. *Heart* 88: 142.
7. Galdzicka, M., et al. 2002. A new gene, EVC2, is mutated in Ellis-van Creveld syndrome. *Mol. Genet. Metab.* 77: 291-295.
8. Ruiz-Perez, V.L., et al. 2003. Mutations in two nonhomologous genes in a head-to-head configuration cause Ellis-van Creveld syndrome. *Am. J. Hum. Genet.* 72: 728-732.

CHROMOSOMAL LOCATION

Genetic locus: EVC2 (human) mapping to 4p16.2; Evc2 (mouse) mapping to 5 B3.

SOURCE

EVC2 (F-12) is a mouse monoclonal antibody specific for an epitope mapping between amino acids 57-89 near the N-terminus of EVC2 of mouse origin.

PRODUCT

Each vial contains 200 µg IgM kappa light chain in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-393128 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% stabilizer protein).

RESEARCH USE

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

APPLICATIONS

EVC2 (F-12) is recommended for detection of EVC2 of mouse, rat and human origin by Western Blotting (starting dilution 1:100, 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), 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 EVC2 siRNA (h): sc-105339, EVC2 siRNA (m): sc-144961, EVC2 shRNA Plasmid (h): sc-105339-SH, EVC2 shRNA Plasmid (m): sc-144961-SH, EVC2 shRNA (h) Lentiviral Particles: sc-105339-V and EVC2 shRNA (m) Lentiviral Particles: sc-144961-V.

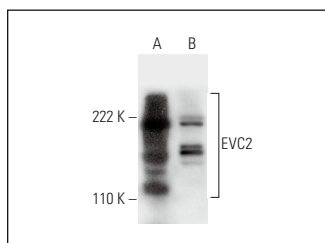
Molecular Weight of EVC2: 153 kDa.

Positive Controls: U-698-M whole cell lysate: sc-364799 or F9 cell lysate: sc-2245.

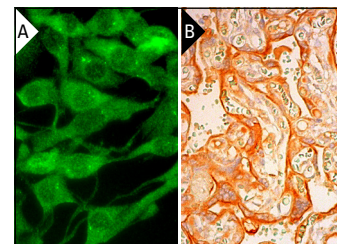
RECOMMENDED SUPPORT REAGENTS

To ensure optimal results, the following support reagents are recommended: 1) Western Blotting: use m-IgGκ BP-HRP: sc-516102 or m-IgGκ BP-HRP (Cruz Marker): sc-516102-CM (dilution range: 1:1000-1:10000), Cruz Marker™ Molecular Weight Standards: sc-2035, UltraCruz® Blocking Reagent: sc-516214 and Western Blotting Luminol Reagent: sc-2048. 2) Immunoprecipitation: use Protein L-Agarose: sc-2336 (0.5 ml agarose/2.0 ml). 3) Immunofluorescence: use m-IgGκ BP-FITC: sc-516140 or m-IgGκ BP-PE: sc-516141 (dilution range: 1:50-1:200) with UltraCruz® Mounting Medium: sc-24941 or UltraCruz® Hard-set Mounting Medium: sc-359850. 4) Immunohistochemistry: use m-IgGκ BP-HRP: sc-516102 with DAB, 50X: sc-24982 and Immunohisto-mount: sc-45086, or Organo/Limonene Mount: sc-45087.

DATA



EVC2 (F-12): sc-393128. Western blot analysis of EVC2 expression in U-698-M (A) and F9 (B) whole cell lysates.



EVC2 (F-12): sc-393128. Immunofluorescence staining of methanol-fixed NIH/3T3 cells showing cytoplasmic and nuclear localization (A). Immunoperoxidase staining of formalin fixed, paraffin-embedded human placenta tissue showing cytoplasmic and membrane staining of trophoblastic cells (B).

STORAGE

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