

EVC2 (L-20): sc-28391

BACKGROUND

Ellis van Creveld syndrome 2 (EVC2), also designated limbin, 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.

SOURCE

EVC2 (L-20) is an affinity purified goat polyclonal antibody raised against a peptide mapping near the C-terminus of EVC2 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.

Blocking peptide available for competition studies, sc-28391 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

STORAGE

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

APPLICATIONS

EVC2 (L-20) is recommended for detection of EVC2 of human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), 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 EVC2 siRNA (h): sc-105339, EVC2 shRNA Plasmid (h): sc-105339-SH and EVC2 shRNA (h) Lentiviral Particles: sc-105339-V.

Positive Controls: MCF7 nuclear extract: sc-2149 or U-698-M whole cell lysate: sc-364799.

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) 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.

SELECT PRODUCT CITATIONS

1. Fleron, M., et al. 2010. Novel post-digest isotope coded protein labeling method for phospho- and glycoproteome analysis. *J. Proteomics* 73: 1986-2005.

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.



Try **EVC2 (F-12): sc-393128**, our highly recommended monoclonal alternative to EVC2 (L-20).