

EVC2 siRNA (h): sc-105339

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. Tompson, S.W., et al. 2001. Ellis-van Creveld syndrome resulting from segmental uniparental disomy of chromosome 4. *J. Med. Genet.* 38: E18.
5. Arya, L., et al. 2001. Ellis-van Creveld syndrome: a report of two cases. *Pediatr. Dermatol.* 18: 485-489.
6. Galdzicka, M., et al. 2002. A new gene, EVC2, is mutated in Ellis-van Creveld syndrome. *Mol. Genet. Metab.* 77: 291-295.
7. Sajeev, C.G., et al. 2002. Images in cardiology: common atrium in a child with Ellis-Van Creveld syndrome. *Heart* 88: 142.
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.

PRODUCT

EVC2 siRNA (h) is a pool of 3 target-specific 19-25 nt siRNAs designed to knock down gene expression. Each vial contains 3.3 nmol of lyophilized siRNA, sufficient for a 10 μ M solution once resuspended using protocol below. Suitable for 50-100 transfections. Also see EVC2 shRNA Plasmid (h): sc-105339-SH and EVC2 shRNA (h) Lentiviral Particles: sc-105339-V as alternate gene silencing products.

For independent verification of EVC2 (h) gene silencing results, we also provide the individual siRNA duplex components. Each is available as 3.3 nmol of lyophilized siRNA. These include: sc-105339A, sc-105339B and sc-105339C.

PROTOCOLS

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

STORAGE AND RESUSPENSION

Store lyophilized siRNA duplex at -20° C with desiccant. Stable for at least one year from the date of shipment. Once resuspended, store at -20° C, avoid contact with RNases and repeated freeze thaw cycles.

Resuspend lyophilized siRNA duplex in 330 μ l of the RNase-free water provided. Resuspension of the siRNA duplex in 330 μ l of RNase-free water makes a 10 μ M solution in a 10 μ M Tris-HCl, pH 8.0, 20 mM NaCl, 1 mM EDTA buffered solution.

APPLICATIONS

EVC2 siRNA (h) is recommended for the inhibition of EVC2 expression in human cells.

SUPPORT REAGENTS

For optimal siRNA transfection efficiency, Santa Cruz Biotechnology's siRNA Transfection Reagent: sc-29528 (0.3 ml), siRNA Transfection Medium: sc-36868 (20 ml) and siRNA Dilution Buffer: sc-29527 (1.5 ml) are recommended. Control siRNAs or Fluorescein Conjugated Control siRNAs are available as 10 μ M in 66 μ l. Each contain a scrambled sequence that will not lead to the specific degradation of any known cellular mRNA. Fluorescein Conjugated Control siRNAs include: sc-36869, sc-44239, sc-44240 and sc-44241. Control siRNAs include: sc-37007, sc-44230, sc-44231, sc-44232, sc-44233, sc-44234, sc-44235, sc-44236, sc-44237 and sc-44238.

GENE EXPRESSION MONITORING

EVC2 (F-12): sc-393128 is recommended as a control antibody for monitoring of EVC2 gene expression knockdown by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000) or immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500).

To ensure optimal results, the following support reagents are recommended: 1) Western Blotting: use m-IgG κ BP-HRP: sc-516102 or m-IgG κ 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) Immunofluorescence: use m-IgG κ BP-FITC: sc-516140 or m-IgG κ BP-PE: sc-516141 (dilution range: 1:50-1:200) with UltraCruz® Mounting Medium: sc-24941 or UltraCruz® Hard-set Mounting Medium: sc-359850.

RT-PCR REAGENTS

Semi-quantitative RT-PCR may be performed to monitor EVC2 gene expression knockdown using RT-PCR Primer: EVC2 (h)-PR: sc-105339-PR (20 μ l). Annealing temperature for the primers should be 55-60° C and the extension temperature should be 68-72° C.

RESEARCH USE

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