

EVC siRNA (h): sc-72235

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., et al. 1996. The gene for the Ellis-van Creveld syndrome is located on chromosome 4p16. *Genomics* 35: 1-5.
2. Ruiz-Perez, V.L., et al. 2000. Mutations in a new gene in Ellis-van Creveld syndrome and Weyers acrodistal dysostosis. *Nat. Genet.* 24: 283-286.
3. Galdzicka, M., et al. 2002. A new gene, EVC is mutated in Ellis-van Creveld syndrome. *Mol. Genet. Metab.* 77: 291-295.
4. Mostafa, M.I., et al. 2005. Unusual pattern of inheritance and orodental changes in the Ellis-van Creveld syndrome. *Genet. Couns.* 16: 75-83.
5. Scurlock, D., et al. 2005. Ellis-van Creveld syndrome and dyserythropoiesis. *Arch. Pathol. Lab. Med.* 129: 680-682.
6. van Hagen, J.M., et al. 2005. From gene to disease; EVC, EVC2, and Ellis-van Creveld syndrome. *Ned. Tijdschr. Geneesk.* 149: 929-931.
7. Ye, X., et al. 2006. A novel heterozygous deletion in the EVC2 gene causes Weyers acrofacial dysostosis. *Hum. Genet.* 119: 199-205.
8. Ruiz-Perez, V.L., et al. 2007. Evc is a positive mediator of Ihh-regulated bone growth that localises at the base of chondrocyte cilia. *Development* 134: 2903-2912.

CHROMOSOMAL LOCATION

Genetic locus: EVC (human) mapping to 4p16.2.

PRODUCT

EVC 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 EVC shRNA Plasmid (h): sc-72235-SH and EVC shRNA (h) Lentiviral Particles: sc-72235-V as alternate gene silencing products.

For independent verification of EVC (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-72235A, sc-72235B and sc-72235C.

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

EVC siRNA (h) is recommended for the inhibition of EVC 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

EVC (H-1): sc-271955 is recommended as a control antibody for monitoring of EVC 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) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml). 3) 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 EVC gene expression knockdown using RT-PCR Primer: EVC (h)-PR: sc-72235-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.