



SZABO SCANDIC

Part of Europa Biosite

Produktinformation



Forschungsprodukte & Biochemikalien



Zellkultur & Verbrauchsmaterial



Diagnostik & molekulare Diagnostik



Laborgeräte & Service

Weitere Information auf den folgenden Seiten!
See the following pages for more information!



Lieferung & Zahlungsart

siehe unsere [Liefer- und Versandbedingungen](#)

Zuschläge

- Mindermengenzuschlag
- Trockeneiszuschlag
- Gefahrgutzuschlag
- Expressversand

SZABO-SCANDIC HandelsgmbH

Quellenstraße 110, A-1100 Wien

T. +43(0)1 489 3961-0

F. +43(0)1 489 3961-7

mail@szabo-scandic.com

www.szabo-scandic.com

[linkedin.com/company/szaboscandic](https://www.linkedin.com/company/szaboscandic) 

EVC (m): 293T Lysate: sc-120134

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 acrodermal 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., Meitingner, T., Polymeropoulos, M.H. and Goodship, J. 2000. Mutations in a new gene in Ellis-van Creveld syndrome and Weyers acrodermal 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 orodental 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. Scurlock, D., Ostler, D., Nguyen, A. and Wahed, A. 2005. Ellis-van Creveld syndrome and dyserythropoiesis. *Arch. Pathol. Lab. Med.* 129: 680-682.

CHROMOSOMAL LOCATION

Genetic locus: Evc (mouse) mapping to 5 B3.

PRODUCT

EVC (m): 293T Lysate represents a lysate of mouse EVC transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

STORAGE

Store at -20° C. Repeated freezing and thawing should be minimized. Sample vial should be boiled once prior to use. Non-hazardous. No MSDS required.

PROTOCOLS

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

APPLICATIONS

EVC (m): 293T Lysate is suitable as a Western Blotting positive control for mouse reactive EVC antibodies. Recommended use: 10-20 µl per lane.

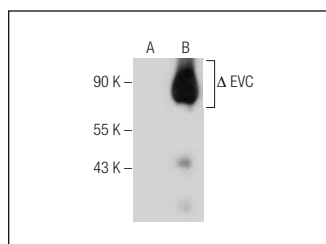
Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

EVC (C-6): sc-377157 is recommended as a positive control antibody for Western Blot analysis of enhanced mouse EVC expression in EVC transfected 293T cells (starting dilution 1:100, dilution range 1:100-1:1,000).

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.

DATA



EVC (C-6): sc-377157. Western blot analysis of EVC expression in non-transfected: sc-117752 (A) and truncated mouse EVC transfected: sc-120134 (B) 293T whole cell lysates.

RESEARCH USE

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