

cadherin-23 (h): 293T Lysate: sc-112533

BACKGROUND

Cadherin-23 represents the first in this family of calcium binding proteins of which mutations in the extracellular calcium binding domain contribute to an inherited disorder, Usher syndrome type 1D (USH1D). Patients with USH1D exhibit congenital sensorineural hearing loss, vestibular dysfunction and visual impairment due to early onset of retinitis pigmentosa (RP). In the inner ear, cadherin-23 interacts with Myosin VIIA and Harmonin to form a functional network during hair cell differentiation and in the retina to assemble a supra-molecular complex contributing to the organization of the cytoskeletal matrices of the pre- and post-synaptic region. A number of cadherin-23 splice variants exist in association with various phenotypic expression, indicating that differential mutations result in variable presentation of the disease.

REFERENCES

1. Di Palma, F., Pellegrino, R. and Noben-Trauth, K. 2001. Genomic structure, alternative splice forms and normal and mutant alleles of cadherin 23 (Cdh23). *Gene* 281: 31-41.
2. Bolz, H., von Brederlow, B., Ramirez, A., Bryda, E.C., Kutsche, K., Nothwang, H.G., Seeliger, M., del C-Salcedo Cabrera, M., Vila, M.C., Molina, O.P., Gal, A. and Kubisch, C. 2001. Mutation of CDH23, encoding a new member of the cadherin gene family, causes Usher syndrome type 1D. *Nat. Genet.* 27: 108-112.
3. Boëda, B., El-Amraoui, A., Bahloul, A., Goodyear, R., Daviet, L., Blanchard, S., Perfettini, I., Fath, K.R., Shorte, S., Reiners, J., Houdusse, A., Legrain, P., Wolfrum, U., Richardson, G. and Petit, C. 2002. Myosin VIIa, Harmonin and cadherin 23, three Usher I gene products that cooperate to shape the sensory hair cell bundle. *EMBO J.* 21: 6689-6699.
4. Siemens, J., Kazmierczak, P., Reynolds, A., Sticker, M., Littlewood-Evans, A. and Müller, U. 2002. The Usher syndrome proteins cadherin 23 and Harmonin form a complex by means of PDZ-domain interactions. *Proc. Natl. Acad. Sci. USA* 99: 14946-14951.
5. de Brouwer, A.P., Pennings, R.J., Roeters, M., Van Hauwe, P., Astuto, L.M., Hoefsloot, L.H., Huygen, P.L., van den Helm, B., Deutman, A.F. M., Bork, J., Kimberling, W.J., Cremers, F.P., Cremers, C.W. and Kremer, H. 2003. Mutations in the calcium-binding motifs of CDH23 and the 35delG mutation in GJB2 cause hearing loss in one family. *Hum. Genet.* 112: 156-163.
6. Noben-Trauth, K., Zheng, Q.Y. and Johnson, K.R. 2003. Association of cadherin 23 with polygenic inheritance and genetic modification of sensorineural hearing loss. *Nat. Genet.* 35: 21-23.
7. Reiners, J., Reidel, B., El-Amraoui, A., Boëda, B., Huber, I., Petit, C. and Wolfrum, U. 2003. Differential distribution of Harmonin isoforms and their possible role in Usher-1 protein complexes in mammalian photoreceptor cells. *Invest. Ophthalmol. Vis. Sci.* 44: 5006-5015.
8. Siemens, J., Lillo, C., Dumont, R.A., Reynolds, A., Williams, D.S., Gillespie, P.G. and Müller, U. 2004. Cadherin 23 is a component of the tip link in hair-cell stereocilia. *Nature* 428: 950-955.

CHROMOSOMAL LOCATION

Genetic locus: CDH23 (human) mapping to 10q22.1.

PRODUCT

cadherin-23 (h): 293T Lysate represents a lysate of human cadherin-23 transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

APPLICATIONS

cadherin-23 (h): 293T Lysate is suitable as a Western Blotting positive control for human reactive cadherin-23 antibodies. Recommended use: 10-20 µl per lane.

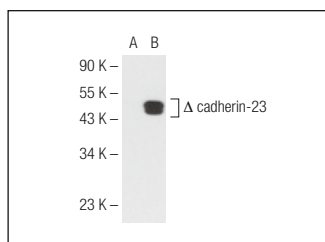
Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

cadherin-23 (H-7): sc-166066 is recommended as a positive control antibody for Western Blot analysis of enhanced human cadherin-23 expression in cadherin-23 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



cadherin-23 (H-7): sc-166066. Western blot analysis of cadherin-23 expression in non-transfected: sc-117752 (A) and truncated human cadherin-23 transfected: sc-112533 (B) 293T whole cell lysates.

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.

RESEARCH USE

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

PROTOCOLS

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