



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) 

TMCC3 (m): 293T Lysate: sc-124089

BACKGROUND

TMCC3 (transmembrane and coiled-coil domains protein 3) is a 477 amino acid multi-pass membrane protein that belongs to the TEX28 family. The gene that encodes TMCC3 contains approximately 83,429 bases and maps to human chromosome 12q22. Encoding over 1,100 genes within 132 million bases, chromosome 12 makes up about 4.5% of the human genome. A number of skeletal deformities are linked to chromosome 12, including hypochondrogenesis, achondrogenesis and Kniest dysplasia. Noonan syndrome, which includes heart and facial developmental defects among the primary symptoms, is caused by a mutant form of PTPN11 gene product, SH-PTP2. Chromosome 12 is also home to a homeobox gene cluster, which encodes crucial transcription factors for morphogenesis, and the natural killer complex gene cluster, encoding C-type lectin proteins which mediate the NK cell response to MHC I interaction. Trisomy 12p leads to facial development defects, seizure disorders and a host of other symptoms which vary in severity depending on the extent of mosaicism. It is most severe in cases of complete trisomy.

REFERENCES

- Allen, T.L., Brothman, A.R., Carey, J.C. and Chance, P.F. 1996. Cytogenetic and molecular analysis in trisomy 12p. *Am. J. Med. Genet.* 63: 250-256.
- Delgado Carrasco, J., Casanova Morcillo, A., Zabalza Alvillos, M. and Ayala Garcés, A. 2001. Achondrogenesis type II-hypochondrogenesis: radiological features. Case report. *An. Esp. Pediatr.* 55: 553-557.
- Yokoyama, T., Nakatani, S. and Murakami, A. 2003. A case of Kniest dysplasia with retinal detachment and the mutation analysis. *Am. J. Ophthalmol.* 136: 1186-1188.
- Forzano, F., Lituania, M., Viassolo, A., Superti-Furga, V., Wildhardt, G., Zabel, B. and Faravelli, F. 2007. A familial case of achondrogenesis type II caused by a dominant COL2A1 mutation and "patchy" expression in the mosaic father. *Am. J. Med. Genet. A* 143A: 2815-2820.
- Wainwright, H. and Beighton, P. 2008. Visceral manifestations of hypochondrogenesis. *Virchows Arch.* 453: 203-207.
- Lo, F.S., Luo, J.D., Lee, Y.J., Shu, S.G., Kuo, M.T. and Chiou, C.C. 2009. High resolution melting analysis for mutation detection for PTPN11 gene: applications of this method for diagnosis of Noonan syndrome. *Clin. Chim. Acta* 409: 75-77.
- Benussi, D.G., Costa, P., Zollino, M., Murdolo, M., Petix, V., Carrozzi, M. and Pecile, V. 2009. Trisomy 12p and monosomy 4p: phenotype-genotype correlation. *Genet. Test. Mol. Biomarkers* 13: 199-204.

CHROMOSOMAL LOCATION

Genetic locus: *Tmcc3* (mouse) mapping to 10 C2.

PRODUCT

TMCC3 (m): 293T Lysate represents a lysate of mouse TMCC3 transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

APPLICATIONS

TMCC3 (m): 293T Lysate is suitable as a Western Blotting positive control for mouse reactive TMCC3 antibodies. Recommended use: 10-20 µl per lane.

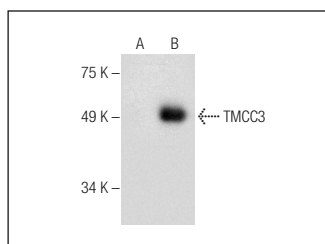
Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

TMCC3 (F-2): sc-390162 is recommended as a positive control antibody for Western Blot analysis of enhanced mouse TMCC3 expression in TMCC3 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



TMCC3 (F-2): sc-390162. Western blot analysis of TMCC3 expression in non-transfected: sc-117752 (A) and mouse TMCC3 transfected: sc-124089 (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.