



# TMC3 (N-16): sc-248834

## BACKGROUND

TMC3 (transmembrane channel-like protein 3) is a 1,100 amino acid multi-pass membrane protein that belongs to the TMC family and exists as 2 alternatively spliced isoforms. The gene that encodes TMC3 consists of nearly 42,000 bases and maps to human chromosome 15q25.1. Housing approximately 106 million base pairs and encoding more than 700 genes, chromosome 15 makes up about 3% of the human genome. Angelman and Prader-Willi syndromes are associated with loss of function or deletion of genes in the 15q11-q13 region. In the case of Angelman syndrome, this loss is due to inactivity of the maternal 15q11-q13 encoded UBE3A gene in the brain by either chromosomal deletion or mutation. In cases of Prader-Willi syndrome, there is a partial or complete deletion of this region from the paternal copy of chromosome 15. Tay-Sachs disease is a lethal disorder associated with mutations of the HEXA gene, which is encoded by chromosome 15. Marfan syndrome is associated with chromosome 15 through the FBN1 gene.

## REFERENCES

- Hurowitz, G.I., Silver, J.M., Brin, M.F., Williams, D.T. and Johnson, W.G. 1993. Neuropsychiatric aspects of adult-onset Tay-Sachs disease: two case reports with several new findings. *J. Neuropsychiatry Clin. Neurosci.* 5: 30-36.
- Keresztes, G., Mutai, H. and Heller, S. 2003. TMC and EVER genes belong to a larger novel family, the TMC gene family encoding transmembrane proteins. *BMC Genomics* 4: 24.
- Kurima, K., Yang, Y., Sorber, K. and Griffith, A.J. 2003. Characterization of the transmembrane channel-like (TMC) gene family: functional clues from hearing loss and epidermodysplasia verruciformis. *Genomics* 82: 300-308.
- Midla, G.S. 2008. Diagnosis and management of patients with Marfan syndrome. *JAAPA* 21: 21-25.
- Dan, B. 2009. Angelman syndrome: current understanding and research prospects. *Epilepsia* 50: 2331-2339.
- Ferrer-Bolufer, I., Dalmau, J., Quiroga, R., Oltra, S., Orellana, C., Monfort, S., Roselló, M., De La Osa, A. and Martinez, F. 2009. Tyrosinemia type 1 and Angelman syndrome due to paternal uniparental isodisomy 15. *J. Inherit. Metab. Dis.* 32: S349-S353.
- Wawrzik, M., Unmehopa, U.A., Swaab, D.F., van de Nes, J., Buiting, K. and Horsthemke, B. 2010. The C15orf2 gene in the Prader-Willi syndrome region is subject to genomic imprinting and positive selection. *Neurogenetics* 11: 153-161.

## CHROMOSOMAL LOCATION

Genetic locus: TMC3 (human) mapping to 15q25.1; Tmc3 (mouse) mapping to 7 D3.

## SOURCE

TMC3 (N-16) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an N-terminal cytoplasmic domain of TMC3 of human origin.

## PRODUCT

Each vial contains 200 µg IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-248834 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

## APPLICATIONS

TMC3 (N-16) is recommended for detection of TMC3 of mouse, rat and human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000); non cross-reactive with other TMC family members.

TMC3 (N-16) is also recommended for detection of TMC3 in additional species, including equine, canine and porcine.

Suitable for use as control antibody for TMC3 siRNA (h): sc-90001, TMC3 siRNA (m): sc-154316, TMC3 shRNA Plasmid (h): sc-90001-SH, TMC3 shRNA Plasmid (m): sc-154316-SH, TMC3 shRNA (h) Lentiviral Particles: sc-90001-V and TMC3 shRNA (m) Lentiviral Particles: sc-154316-V.

Molecular Weight of TMC3 isoforms: 126/83 kDa.

## RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use donkey anti-goat IgG-HRP: sc-2020 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible donkey anti-goat IgG-HRP: sc-2033 (dilution range: 1:2000-1:5000), Cruz Marker™ Molecular Weight Standards: sc-2035, TBS Blotto A Blocking Reagent: sc-2333 and Western Blotting Luminol Reagent: sc-2048. 2) Immunofluorescence: use donkey anti-goat IgG-FITC: sc-2024 (dilution range: 1:100-1:400) or donkey anti-goat IgG-TR: sc-2783 (dilution range: 1:100-1:400) with UltraCruz™ Mounting Medium: sc-24941.

## STORAGE

Store at 4° C, **\*\*DO NOT FREEZE\*\***. Stable for one year from the date of shipment. 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](http://www.scbt.com) or our catalog for detailed protocols and support products.