

# TMEM248 (E-17): sc-241324

## BACKGROUND

TMEM248 (transmembrane protein 248) is a 314 amino acid multi-pass membrane protein. Existing as two alternatively spliced isoforms, TMEM248 is encoded by a gene that maps to human chromosome 7q11.21 and mouse chromosome 5 G1.3. Chromosome 7 is about 158 million bases long, encodes over 1,000 genes and makes up about 5% of the human genome. Chromosome 7 has been linked to osteogenesis imperfecta, Pendred syndrome, lissencephaly, citrullinemia and Shwachman-Diamond syndrome. The deletion of a portion of the q arm of chromosome 7 is associated with Williams-Beuren syndrome, a condition characterized by mild mental retardation, an unusual comfort and friendliness with strangers and an elfin appearance. Deletions of portions of the q arm of chromosome 7 are also seen in a number of myeloid disorders including cases of acute myelogenous leukemia and myelodysplasia.

## REFERENCES

1. Tspouras, P., et al. 1983. Restriction fragment length polymorphism associated with the pro  $\alpha$  2(I) gene of human type I procollagen. Application to a family with an autosomal dominant form of osteogenesis imperfecta. *J. Clin. Invest.* 72: 1262-1267.
2. Liang, H., et al. 1998. Molecular anatomy of chromosome 7q deletions in myeloid neoplasms: evidence for multiple critical loci. *Proc. Natl. Acad. Sci. USA* 95: 3781-3785.
3. Hillier, L.W., et al. 2003. The DNA sequence of human chromosome 7. *Nature* 424: 157-164.
4. Eckert, M.A., et al. 2006. The neurobiology of Williams syndrome: cascading influences of visual system impairment? *Cell. Mol. Life Sci.* 63: 1867-1875.
5. Osborne, L.R., et al. 2006. Williams-Beuren syndrome diagnosis using fluorescence *in situ* hybridization. *Methods Mol. Med.* 126:113-128.
6. Reiner, O., et al. 2006. Lissencephaly 1 linking to multiple diseases: mental retardation, neurodegeneration, schizophrenia, male sterility, and more. *Neuromolecular Med.* 8: 547-565.

## CHROMOSOMAL LOCATION

Genetic locus: TMEM248 (human) mapping to 7q11.21; Tmem248 (mouse) mapping to 5 G1.3.

## SOURCE

TMEM248 (E-17) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of TMEM248 of human origin.

## PRODUCT

Each vial contains 200  $\mu$ g IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-241324 P, (100  $\mu$ g peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

## APPLICATIONS

TMEM248 (E-17) is recommended for detection of TMEM248 of mouse, rat and human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunoprecipitation [1-2  $\mu$ g per 100-500  $\mu$ g of total protein (1 ml of cell lysate)], 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).

TMEM248 (E-17) is also recommended for detection of TMEM248 in additional species, including equine, canine, bovine and porcine.

Suitable for use as control antibody for TMEM248 siRNA (h): sc-89672, TMEM248 siRNA (m): sc-108102, TMEM248 shRNA Plasmid (h): sc-89672-SH, TMEM248 shRNA Plasmid (m): sc-108102-SH, TMEM248 shRNA (h) Lentiviral Particles: sc-89672-V and TMEM248 shRNA (m) Lentiviral Particles: sc-108102-V.

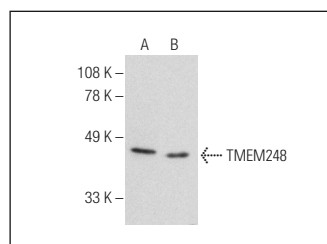
Molecular Weight of TMEM248 isoforms: 35/22 kDa.

Positive Controls: MCF7 whole cell lysate: sc-2206 or HeLa whole cell lysate: sc-2200.

## 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) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml). 3) 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.

## DATA



TMEM248 (E-17): sc-241324. Western blot analysis of TMEM248 expression in MCF7 (A) and HeLa (B) whole cell lysates.

## 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.