

TMEM247 (S-19): sc-247650

BACKGROUND

TMEM247 is a 219 amino acid multi-pass membrane protein that maps to human chromosome 2. Human chromosome 2 consists of 237 million bases, encodes over 1,400 genes and makes up approximately 8% of the human genome. A number of genetic diseases are linked to genes on chromosome 2. Harlequin ichthyosis, a rare and morbid skin deformity, is associated with mutations in the ABCA12 gene. The lipid metabolic disorder sitosterolemia is associated with ABCG5 and ABCG8. An extremely rare recessive genetic disorder, Alström syndrome, is due to mutations in the ALMS1 gene. Interestingly, chromosome 2 contains what appears to be a vestigial second centromere and vestigial telomeres which gives credence to the hypothesis that human chromosome 2 is the result of an ancient fusion of two ancestral chromosomes seen in modern form today in apes.

REFERENCES

- Baldini, A., et al. 1993. An alphoid DNA sequence conserved in all human and great ape chromosomes: evidence for ancient centromeric sequences at human chromosomal regions 2q21 and 9q13. *Hum. Genet.* 90: 577-583.
- Patel, S.B., et al. 1998. Mapping a gene involved in regulating dietary cholesterol absorption. The sitosterolemia locus is found at chromosome 2p21. *J. Clin. Invest.* 102: 1041-1044.
- Zumsteg, U., et al. 2000. Alstrom syndrome: confirmation of linkage to chromosome 2p12-13 and phenotypic heterogeneity in three affected sibs. *J. Med. Genet.* 37: E8.
- Shulenin, S., et al. 2001. An ATP-binding cassette gene (ABCG5) from the ABCG (White) gene subfamily maps to human chromosome 2p21 in the region of the *Sitosterolemia locus*. *Cytogenet. Cell Genet.* 92: 204-208.
- Hearn, T., et al. 2002. Mutation of ALMS1, a large gene with a tandem repeat encoding 47 amino acids, causes Alström syndrome. *Nat. Genet.* 31: 79-83.
- Kelsell, D.P., et al. 2005. Mutations in ABCA12 underlie the severe congenital skin disease harlequin ichthyosis. *Am. J. Hum. Genet.* 76: 794-803.
- Horvath, J.E., et al. 2005. Punctuated duplication seeding events during the evolution of human chromosome 2p11. *Genome Res.* 15: 914-927.

CHROMOSOMAL LOCATION

Genetic locus: Tmem247 (mouse) mapping to 17 E4.

SOURCE

TMEM247 (S-19) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of TMEM247 of mouse 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-247650 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

TMEM247 (S-19) is recommended for detection of TMEM247 of mouse and rat 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).

Suitable for use as control antibody for TMEM247 siRNA (m): sc-108499, TMEM247 shRNA Plasmid (m): sc-108499-SH and TMEM247 shRNA (m) Lentiviral Particles: sc-108499-V.

Molecular Weight of TMEM247: 25 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 or our catalog for detailed protocols and support products.