SANTA CRUZ BIOTECHNOLOGY, INC.

FAM179A (E-19): sc-246647

SANTA CRUZ BIOTECHNOLOGY

The Power to Question

BACKGROUND

FAM179A is a 1,019 amino acid protein that is expressed as 3 alternatively spliced isoforms. The gene encoding FAM179A maps to human chromosome 2, the second largest chromosome that consists of 237 million bases encoding over 1,400 genes and making up approximately 8% of the human genome. A number of genetic diseases are linked to genes on chromosome 2. Harlequin icthyosis, 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

1. IJdo, J.W., Baldini, A., Ward, D.C., Reeders, S.T. and Wells, R.A. 1991. Origin of human chromosome 2: an ancestral telomere-telomere fusion. Proc. Natl. Acad. Sci. USA 88: 9051-9055.
2. Avarello, R., Pedicini, A., Caiulo, A., Zuffardi, O. and Fraccaro, M. 1992. Evidence for an ancestral alphoid domain on the long arm of human chromosome 2. Hum. Genet. 89: 247-249.
3. Zumsteg, U., Muller, P.Y. and Miserez, A.R. 2000. Alstrom syndrome: confirmation of linkage to chromosome 2p12-13 and phenotypic heterogeneity in three affected sibs. J. Med. Genet. 37: E8.
4. Hillier, L.W., Graves, T.A., Fulton, R.S., Fulton, L.A., Pepin, K.H., Minx, P., Wagner-McPherson, C., Layman, D., Wylie, K., Sekhon, M., Becker, M.C., Fewell, G.A., et al. 2005. Generation and annotation of the DNA sequences of human chromosomes 2 and 4. Nature 434: 724-731.
5. Thomas, A.C., Cullup, T., Norgett, E.E., Hill, T., Barton, S., Dale, B.A., Sprecher, E., Sheridan, E., Taylor, A.E., Wilroy, R.S., DeLozier, C., Burrows, N., Goodyear, H., Fleckman, P., et al. 2006. ABCA12 is the major harlequin ichthyosis gene. J. Invest. Dermatol. 126: 2408-2413.
6. Tabas, I. 2007. A two-carbon switch to sterol-induced autophagic death. Autophagy 3: 38-41.
7. Akiyama, M., Sakai, K., Sato, T., McMillan, J.R., Goto, M., Sawamura, D. and Shimizu, H. 2007. Compound heterozygous ABCA12 mutations including a novel nonsense mutation underlie harlequin ichthyosis. Dermatology 215: 155-159.
8. Marshall, J.D., Hinman, E.G., Collin, G.B., Beck, S., Cerqueira, R., Maffei, P., Milan, G., Zhang, W., Wilson, D.I., Hearn, T., Tavares, P., Vettor, R., Veronese, C., Martin, M., So, W.V., Nishina, P.M. and Naggert, J.K. 2007. Spectrum of ALMS1 variants and evaluation of genotype-phenotype correlations in Alström syndrome. Hum. Mutat. 28: 1114-1123.

CHROMOSOMAL LOCATION

Genetic locus: FAM179A (human) mapping to 2p23.2; Fam179a (mouse) mapping to 17 E1.3.

SOURCE

FAM179A (E-19) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of FAM179A 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-246647 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

FAM179A (E-19) is recommended for detection of FAM179A of mouse 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 FAM179B.

FAM179A (E-19) is also recommended for detection of FAM179A in additional species, including porcine.

Suitable for use as control antibody for FAM179A siRNA (h): sc-94363, FAM179A siRNA (m): sc-108947, FAM179A shRNA Plasmid (h): sc-94363-SH, FAM179A shRNA Plasmid (m): sc-108947-SH, FAM179A shRNA (h) Lentiviral Particles: sc-94363-V and FAM179A shRNA (m) Lentiviral Particles: sc-108947-V.

Molecular Weight of FAM179A isoforms 1/2/3: 11/60/27 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.