

# FAM49A (S-15): sc-164335

## BACKGROUND

FAM49A (family with sequence similarity 49, member A) is a 323 amino acid protein that is encoded by a gene which maps to human chromosome 2. The second largest human chromosome, chromosome 2 encodes over 1,400 genes and comprises nearly 8% of the human genome, housing a number of disease-associated genes. Harlequin ichthyosis, a rare and morbid skin deformity, is associated with mutations in the ABCA12 gene, while the lipid metabolic disorder sitosterolemia is associated with defects in the ABCG5 and ABCG8 genes. Additionally, an extremely rare recessive genetic disorder, Alström syndrome, is caused by mutations in the ALMS1 gene, which maps to chromosome 2.

## REFERENCES

- 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.
- 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., Delehaunty, K.D., Miner, T.L., Nash, W.E., et al. 2005. Generation and annotation of the DNA sequences of human chromosomes 2 and 4. *Nature* 434: 724-731.
- 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., Stephens, K.G., Mehta, L., et al. 2006. ABCA12 is the major harlequin ichthyosis gene. *J. Invest. Dermatol.* 126: 2408-2413.
- 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.
- Marshall, J.D., Beck, S., Maffei, P. and Naggert, J.K. 2007. Alström syndrome. *Eur. J. Hum. Genet.* 15: 1193-1202.
- 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.
- Tabas, I. 2007. A two-carbon switch to sterol-induced autophagic death. *Autophagy* 3: 38-41.
- Wang, D.Q. 2007. Regulation of intestinal cholesterol absorption. *Annu. Rev. Physiol.* 69: 221-248.

## CHROMOSOMAL LOCATION

Genetic locus: FAM49A (human) mapping to 2p24.2; Fam49a (mouse) mapping to 12 A1.1.

## SOURCE

FAM49A (S-15) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of FAM49A 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-164335 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

## APPLICATIONS

FAM49A (S-15) is recommended for detection of FAM49A 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 FAM49B.

FAM49A (S-15) is also recommended for detection of FAM49A in additional species, including equine, canine, bovine, porcine and avian.

Suitable for use as control antibody for FAM49A siRNA (h): sc-94318, FAM49A siRNA (m): sc-145045, FAM49A shRNA Plasmid (h): sc-94318-SH, FAM49A shRNA Plasmid (m): sc-145045-SH, FAM49A shRNA (h) Lentiviral Particles: sc-94318-V and FAM49A shRNA (m) Lentiviral Particles: sc-145045-V.

Molecular Weight of FAM49A: 37 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.