SANTA CRUZ BIOTECHNOLOGY, INC.

FLJ34931 (V-12): sc-136690

BACKGROUND

The second largest human chromosome, 2 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. The FLJ34931 gene product has been provisionally designated FLJ34931 pending further characterization.

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. 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., Kremitzki, C., Oddy, L., Du, H., Sun, H., Bradshaw-Cordum, H., Ali, J., Carter, J., Cordes, M., Harris, A., Isak, A., van Brunt, A., Nguyen, C., et al. 2005. Generation and annotation of the DNA sequences of human chromosomes 2 and 4. Nature 434: 724-731.
4. 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., Watson, R.M., Graham, R., Wolf, R., Slavotinek, A., Martin, M., Bourn, D., Mein, C.A., O'Toole, E.A. and Kelsell, D.P. 2006. ABCA12 is the major harlequin ichthyosis gene. J. Invest. Dermatol. 126: 2408-2413.
5. 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.
6. Marshall, J.D., Beck, S., Maffei, P. and Naggert, J.K. 2007. Alström syndrome. Eur. J. Hum. Genet. 15: 1193-1202.
7. 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.
8. Tabas, I. 2007. A two-carbon switch to sterol-induced autophagic death. Autophagy 3: 38-41.
9. Wang, D.Q. 2007. Regulation of intestinal cholesterol absorption. Annu. Rev. Physiol. 69: 221-248.

CHROMOSOMAL LOCATION

Genetic locus: C2orf71 (human) mapping to 2p23.2; BC027072 (mouse) mapping to 17 E1.3.

SOURCE

FLJ34931 (V-12) is an affinity purified rabbit polyclonal antibody raised against a peptide mapping within an internal region of FLJ34931 of human origin.

PRODUCT

Each vial contains 100 µg IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

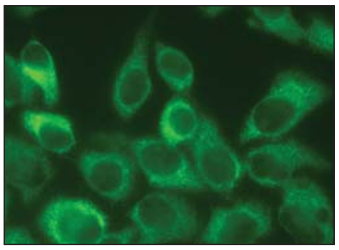
Blocking peptide available for competition studies, sc-136690 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

FLJ34931 (V-12) is recommended for detection of FLJ34931 of mouse, rat and human origin by Western Blotting (starting dilution 1:100, dilution range 1:50-1:500), immunofluorescence (starting dilution 1:25, dilution range 1:25-1:250) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000); non cross-reactive with other FLJ family members.

Suitable for use as control antibody for FLJ34931 siRNA (h): sc-94884, FLJ34931 shRNA Plasmid (h): sc-94884-SH and FLJ34931 shRNA (h) Lentiviral Particles: sc-94884-V.

DATA



FLJ34931 (V-12): sc-136690. Immunofluorescence staining of methanol-fixedHeLa cells showing cytoplasmic localization.

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.