

C2orf76 (Q-18): sc-242184

BACKGROUND

C2orf76 (chromosome 2 open reading frame 76), also known as AIM29 or MGC104437, is a 126 amino acid protein belonging to the UPF0538 family. C2orf76 is encoded by a gene that maps to human chromosome 2q14.2. As the second largest human chromosome, chromosome 2 makes up approximately 8% of the human genome and contains 237 million bases encoding over 1,400 genes. A number of genetic diseases are linked to genes on chromosome 2. Harlequin ichthyosis, a rare 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, Alstrom syndrome, is related to mutations in the ALMS1 gene. Chromosome 2 contains a probable vestigial second centromere as well as vestigial telomeres, which gives credence to the hypothesis that human chromosome 2 formed as a result of an ancient fusion of two ancestral chromosomes, which are still present in modern day 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. 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., 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., et al. 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.L., 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: C2orf76 (human) mapping to 2q14.2; 3110009E18Rik (mouse) mapping to 1 E2.3.

SOURCE

C2orf76 (Q-18) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of C2orf76 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-242184 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

C2orf76 (Q-18) is recommended for detection of C2orf76 of human origin, 3110009E18Rik of mouse origin and the corresponding rat homolog 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).

C2orf76 (Q-18) is also recommended for detection of C2orf76 in additional species, including equine, canine, bovine and porcine.

Suitable for use as control antibody for C2orf76 siRNA (h): sc-94481, 3110009E18Rik siRNA (m): sc-108899, C2orf76 shRNA Plasmid (h): sc-94481-SH, 3110009E18Rik shRNA Plasmid (m): sc-108899-SH, C2orf76 shRNA (h) Lentiviral Particles: sc-94481-V and 3110009E18Rik shRNA (m) Lentiviral Particles: sc-108899-V.

Molecular Weight of C2orf76: 15 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.