# LOC257407 (C-18): sc-247566



The Power to Question

#### **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. It is suggested that chromosome 2 may contain 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 LOC257407 gene product has been provisionally designated LOC257407 pending further characterization.

## **REFERENCES**

- Ijdo, J.W., et al. 1991. Origin of human chromosome 2: an ancestral telomere-telomere fusion. Proc. Natl. Acad. Sci. USA 88: 9051-9055.
- 2. Avarello, R., et al. 1992. Evidence for an ancestral alphoid domain on the long arm of human chromosome 2. Hum. Genet. 89: 247-249.
- Hillier, L.W., et al. 2005. Generation and annotation of the DNA sequences of human chromosomes 2 and 4. Nature 434: 724-731.
- Thomas, A.C., et al. 2006. ABCA12 is the major harlequin ichthyosis gene.
  J. Invest. Dermatol. 126: 2408-2413.
- Akiyama, M., et al. 2007. Compound heterozygous ABCA12 mutations including a novel nonsense mutation underlie harlequin ichthyosis. Dermatology 215: 155-159.
- Marshall, J.D., et al. 2007. Alström syndrome. Eur. J. Hum. Genet. 15: 1193-1202.
- Marshall, J.D., et al. 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: C2orf72 (human) mapping to 2q37.1.

#### SOURCE

LOC257407 (C-18) is an affinity purified goat polyclonal antibody raised against a peptide mapping at the C-terminus of LOC257407 of human origin.

## **PRODUCT**

Each vial contains 200  $\mu g$  lgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-247566 P, (100  $\mu$ g peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

#### **APPLICATIONS**

LOC257407 (C-18) is recommended for detection of LOC257407 of 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).

Suitable for use as control antibody for LOC257407 siRNA (h): sc-94973, LOC257407 shRNA Plasmid (h): sc-94973-SH and LOC257407 shRNA (h) Lentiviral Particles: sc-94973-V.

Molecular Weight of LOC257407: 30 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.

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