

# LOC653186 (V-24): sc-133754

## 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 ichthyosis, 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 LOC653186 gene product has been provisionally designated LOC653186 pending further characterization.

## REFERENCES

1. 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.
3. Hillier, L.W., et al. 2005. Generation and annotation of the DNA sequences of human chromosomes 2 and 4. *Nature* 434: 724-731.
4. Thomas, A.C., et al. 2006. ABCA12 is the major harlequin ichthyosis gene. *J. Invest. Dermatol.* 126: 2408-2413.
5. Akiyama, M., et al. 2007. Compound heterozygous ABCA12 mutations including a novel nonsense mutation underlie harlequin ichthyosis. *Dermatology* 215: 155-159.
6. Marshall, J.D., et al. 2007. Alström syndrome. *Eur. J. Hum. Genet.* 15: 1193-1202.
7. Marshall, J.D., et al. 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.

## CHROMOSOMAL LOCATION

Genetic locus: LOC653186 (human) mapping to 2q11.1.

## SOURCE

LOC653186 (V-24) is a Protein A purified rabbit polyclonal antibody raised against synthetic LOC653186 peptide of human origin.

## PRODUCT

Each vial contains 100 µg IgG in 1.0 ml PBS with < 0.1% sodium azide, 0.1% gelatin and < 0.02% sucrose.

## RESEARCH USE

For research use only, not for use in diagnostic procedures.

## APPLICATIONS

LOC653186 (V-24) is recommended for detection of LOC653186 of human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

Suitable for use as control antibody for LOC653186 siRNA (h): sc-94972, LOC653186 shRNA Plasmid (h): sc-94972-SH and LOC653186 shRNA (h) Lentiviral Particles: sc-94972-V.

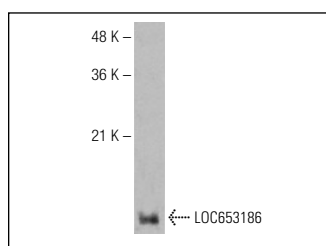
Molecular Weight of LOC653186: 10 kDa.

Positive Controls: Hep G2 cell lysate: sc-2227.

## RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western blotting: use goat anti-rabbit IgG-HRP: sc-2004 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible goat anti-rabbit IgG-HRP: sc-2030 (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.

## DATA



LOC653186 (V-24): sc-133754. Western blot analysis of LOC653186 expression in Hep G2 whole cell lysate.

## STORAGE

Store at 4° C, **\*\*DO NOT FREEZE\*\***. Stable for one year from the date of shipment. Non-hazardous. No MSDS required.

## PROTOCOLS

See our web site at [www.scbt.com](http://www.scbt.com) or our catalog for detailed protocols and support products.