

TEX44 (h): 293T Lysate: sc-175596

BACKGROUND

Chromosome 2, the second largest human chromosome, consists of 237 million bases encoding over 1,400 genes, comprising 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.

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., Krenitzki, 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., 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.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: TEX44 (human) mapping to 2q37.1.

PRODUCT

TEX44 (h): 293T Lysate represents a lysate of human TEX44 transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

STORAGE

Store at -20° C. Repeated freezing and thawing should be minimized. Sample vial should be boiled once prior to use. Non-hazardous. No MSDS required.

APPLICATIONS

TEX44 (h): 293T Lysate is suitable as a Western Blotting positive control for human reactive TEX44 antibodies. Recommended use: 10-20 µl per lane.

Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

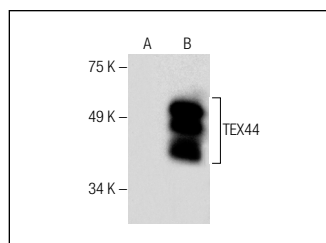
TEX44 (B-1): sc-390142 is recommended as a positive control antibody for Western Blot analysis of enhanced human TEX44 expression in TEX44 transfected 293T cells (starting dilution 1:100, dilution range 1:100-1:1,000).

RECOMMENDED SUPPORT REAGENTS

To ensure optimal results, the following support reagents are recommended:

1) Western Blotting: use m-IgGκ BP-HRP: sc-516102 or m-IgGκ BP-HRP (Cruz Marker): sc-516102-CM (dilution range: 1:1000-1:10000), Cruz Marker™ Molecular Weight Standards: sc-2035, UltraCruz® Blocking Reagent: sc-516214 and Western Blotting Luminol Reagent: sc-2048.

DATA



TEX44 (B-1): sc-390142. Western blot analysis of TEX44 expression in non-transfected: sc-117752 (A) and human TEX44 transfected: sc-175596 (B) 293T whole cell lysates.

RESEARCH USE

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

PROTOCOLS

See our web site at www.scbt.com for detailed protocols and support products.