

dystrotelin (S-20): sc-246495

BACKGROUND

Dystrotelin (DYTN) is a 578 amino acid cell membrane protein that contains one ZZ-type zinc finger. The gene that encodes dystrotelin maps to human chromosome 2, which 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, Alström 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. Jdo, 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., 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., 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., 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.

CHROMOSOMAL LOCATION

Genetic locus: DYTN (human) mapping to 2q33.3.

SOURCE

dystrotelin (S-20) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of dystrotelin of human origin.

STORAGE

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

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-246495 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

dystrotelin (S-20) is recommended for detection of dystrotelin of human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunoprecipitation [1-2 µg per 100-500 µg of total protein (1 ml of cell lysate)], 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 dystrotelin siRNA (h): sc-106948, dystrotelin shRNA Plasmid (h): sc-106948-SH and dystrotelin shRNA (h) Lentiviral Particles: sc-106948-V.

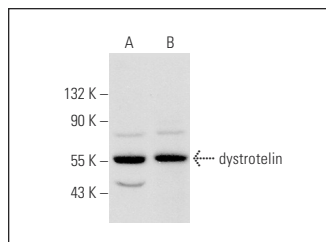
Molecular Weight of dystrotelin: 65 kDa.

Positive Controls: Hep G2 cell lysate: sc-2227 or human liver extract: sc-363766.

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) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml). 3) 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.

DATA



dystrotelin (S-20): sc-246495. Western blot analysis of dystrotelin expression in Hep G2 whole cell lysate (A) and human liver tissue extract (B).

RESEARCH USE

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