

# LOC729774 (A-17): sc-85006

## BACKGROUND

Encoding over 300 genes, chromosome 18 contains about 76 million bases. Trisomy 18, or Edwards syndrome, is the second most common trisomy after Down syndrome. Symptoms of Edwards syndrome include low birth weight, a variety of physical development defects, heart deformations and breathing difficulty. Translocation between chromosome 18 and 14 is the most common translocation in cancers, and occurs in follicular lymphomas. Niemann-Pick disease, hereditary hemorrhagic telangiectasia and erythropoietic protoporphyria are associated with chromosome 18. The TGF $\beta$  modulators, Smad2, Smad4 and Smad7 are encoded by chromosome 18. The LOC729774 gene product has been provisionally designated LOC729774 pending further characterization.

## REFERENCES

- Carstea, E.D., Polymeropoulos, M.H., Parker, C.C., Detera-Wadleigh, S.D., O'Neill, R.R., Patterson, M.C., Goldin, E., Xiao, H., Straub, R.E., Vanier, M.T., et al. 1993. Linkage of Niemann-Pick disease type C to human chromosome 18. *Proc. Natl. Acad. Sci. USA* 90: 2002-2004.
- Petek, E., Pertl, B., Tschernigg, M., Bauer, M., Mayr, J., Wagner, K. and Kroisel, P.M. 2003. Characterisation of a 19-year-old "long-term survivor" with Edwards syndrome. *Genet. Couns.* 14: 239-244.
- Raghavan, S.C., Swanson, P.C., Wu, X., Hsieh, C.L. and Lieber, M.R. 2004. A non-B-DNA structure at the Bcl-2 major breakpoint region is cleaved by the RAG complex. *Nature* 428: 88-93.
- Grosso, S., Pucci, L., Di Bartolo, R.M., Gobbi, G., Bartalini, G., Anichini, C., Scarinci, R., Balestri, M., Farnetani, M.A., Cioni, M., Morgese, G. and Balestri, P. 2005. Chromosome 18 aberrations and epilepsy: a review. *Am. J. Med. Genet. A* 134: 88-94.
- Aurizi, C., Schneider-Yin, X., Sorge, F., Macri, A., Minder, E.I. and Biolcati, G. 2007. Heterogeneity of mutations in the ferrochelatase gene in Italian patients with erythropoietic protoporphyria. *Mol. Genet. Metab.* 90: 402-407.
- Broderick, P., Carvajal-Carmona, L., Pittman, A.M., Webb, E., Howarth, K., Rowan, A., Lubbe, S., Spain, S., Sullivan, K., Fielding, S., Jaeger, E., Vijaykrishnan, J., Kemp, Z., Gorman, M., Chandler, I., Papaemmanuil, E., et al. 2007. A genome-wide association study shows that common alleles of Smad7 influence colorectal cancer risk. *Nat. Genet.* 39: 1315-1317.
- Kamal, A.H. and Prakash, U.B. 2007. Hereditary hemorrhagic telangiectasia. *Mayo Clin. Proc.* 82: 1364.
- Shovlin, C.L., Sulaiman, N.L., Govani, F.S., Jackson, J.E. and Begbie, M.E. 2007. Elevated Factor VIII in hereditary haemorrhagic telangiectasia (HHT): Association with venous thromboembolism. *Thromb. Haemost.* 98: 1031-1039.

## CHROMOSOMAL LOCATION

Genetic locus: LOC729774 (human) mapping to 18p11.21, FAM207A (human) mapping to 21q22.3.

## SOURCE

LOC729774 (A-17) is an affinity purified rabbit polyclonal antibody raised against a peptide mapping within an internal region of LOC729774 of human origin.

## PRODUCT

Each vial contains 100  $\mu$ g IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-85006 P, (100  $\mu$ g peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

## APPLICATIONS

LOC729774 (A-17) is recommended for detection of LOC729774 and C21orf70 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).

LOC729774 (A-17) is also recommended for detection of LOC729774 and C21orf70 in additional species, including equine.

Positive Controls: Human heart extract: sc-363763.

## 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. 2) Immunofluorescence: use goat anti-rabbit IgG-FITC: sc-2012 (dilution range: 1:100-1:400) or goat anti-rabbit IgG-TR: sc-2780 (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](http://www.scbt.com) or our catalog for detailed protocols and support products.