## BACKGROUND

C2orf80 (chromosome 2 open reading frame 80) is a 193 amino acid protein that is encoded by a gene located on human chromosome 2q34. The second largest human chromosome, chromosome 2 consists of 237 million bases, encodes over 1,400 genes and makes 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. 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. D630023F18Rik is the putative mouse homolog of the human C2orf80 protein.

## 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.
9. Wang, D.Q. 2007. Regulation of intestinal cholesterol absorption. Annu. Rev. Physiol. 69: 221-248.

## CHROMOSOMAL LOCATION

Genetic locus: C2orf80 (human) mapping to 2q34.

## SOURCE

C2orf80 (C-18) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of C2orf80 of human origin.

## STORAGE

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

## PRODUCT

Each vial contains $200 \mu \mathrm{glgG}$ in 1.0 ml of PBS with < $0.1 \%$ sodium azide and $0.1 \%$ gelatin.

Blocking peptide available for competition studies, sc-245734 P, (100 $\mu \mathrm{g}$ peptide in 0.5 ml PBS containing $<0.1 \%$ sodium azide and $0.2 \% \mathrm{BSA})$.

## APPLICATIONS

C2orf80 (C-18) is recommended for detection of C2orf80 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).
Molecular Weight of C2orf80: 22 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: 1:1001:400) or donkey anti-goat IgG-TR: sc-2783 (dilution range: 1:100-1:400) with UltraCruz ${ }^{\text {TM }}$ Mounting Medium: sc-24941.

## 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.

