

# CCDC104 (N-16): sc-246063

## BACKGROUND

CCDC104 (coiled-coil domain-containing protein 104) is a 342 amino acid protein that exists as 2 alternatively spliced isoforms. CCDC104 undergoes post-translational phosphorylation following DNA damage, most likely by either ATR or ATM. The gene encoding CCDC104 maps to human chromosome 2, the second largest human chromosome, which 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 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.

## REFERENCES

1. Patel, S.B., et al. 1998. Mapping a gene involved in regulating dietary cholesterol absorption. The sitosterolemia locus is found at chromosome 2p21. *J. Clin. Invest.* 102: 1041-1044.
2. Zumsteg, U., et al. 2000. Alstrom syndrome: confirmation of linkage to chromosome 2p12-13 and phenotypic heterogeneity in three affected sibs. *J. Med. Genet.* 37: E8.
3. Shulenin, S., et al. 2001. An ATP-binding cassette gene (ABCG5) from the ABCG (White) gene subfamily maps to human chromosome 2p21 in the region of the Sitosterolemia locus. *Cytogenet. Cell Genet.* 92: 204-208.
4. Hearn, T., et al. 2002. Mutation of ALMS1, a large gene with a tandem repeat encoding 47 amino acids, causes Alström syndrome. *Nat. Genet.* 31: 79-83.
5. Kelsell, D.P., et al. 2005. Mutations in ABCA12 underlie the severe congenital skin disease harlequin ichthyosis. *Am. J. Hum. Genet.* 76: 794-803.
6. Hillier, L.W., et al. 2005. Generation and annotation of the DNA sequences of human chromosomes 2 and 4. *Nature* 434: 724-731.

## CHROMOSOMAL LOCATION

Genetic locus: CCDC104 (human) mapping to 2p16.1; Ccdc104 (mouse) mapping to 11 A3.3.

## SOURCE

CCDC104 (N-16) is an affinity purified goat polyclonal antibody raised against a peptide mapping near the N-terminus of CCDC104 of human origin.

## 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-246063 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

## STORAGE

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

## APPLICATIONS

CCDC104 (N-16) is recommended for detection of CCDC104 of mouse, rat and 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); non cross-reactive with other CCDC family members.

CCDC104 (N-16) is also recommended for detection of CCDC104 in additional species, including equine, canine, bovine and porcine.

Suitable for use as control antibody for CCDC104 siRNA (h): sc-94606, CCDC104 siRNA (m): sc-142047, CCDC104 shRNA Plasmid (h): sc-94606-SH, CCDC104 shRNA Plasmid (m): sc-142047-SH, CCDC104 shRNA (h) Lentiviral Particles: sc-94606-V and CCDC104 shRNA (m) Lentiviral Particles: sc-142047-V.

Molecular Weight of CCDC104: 39 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: 1:100-1:400) or donkey anti-goat IgG-TR: sc-2783 (dilution range: 1:100-1:400) with UltraCruz™ 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](http://www.scbt.com) or our catalog for detailed protocols and support products.