

# CCDC109A (E-16): sc-246072

## BACKGROUND

CCDC109A (coiled-coil domain containing 109A), also known as MCU (mitochondrial calcium uniporter), is a 351 amino acid mitochondrial multi-pass membrane protein that belongs to the MCU family. Functioning as a calcium transporter, CCDC109A exists as a homooligomer that interacts with CBARA1 (calcium binding atopy-related autoantigen 1). CCDC109A exists as 3 alternatively spliced isoforms and is encoded by a gene that maps to human chromosome 10, which contains over 800 genes and 135 million nucleotides, making up nearly 4.5% of the human genome. PTEN is an important tumor suppressor gene located on chromosome 10 and, when defective, causes a genetic predisposition to cancer development known as Cowden syndrome. The chromosome 10 encoded gene ERCC6 is important for DNA repair and is linked to Cockayne syndrome which is characterized by extreme photosensitivity and premature aging. Tetrahydrobiopterin deficiency and a number of syndromes involving defective skull and facial bone fusion are also linked to chromosome 10. As with most trisomies, trisomy 10 is rare and is deleterious.

## REFERENCES

1. Troelstra, C., et al. 1992. Localization of the nucleotide excision repair gene ERCC6 to human chromosome 10q11-q21. *Genomics* 12: 745-749.
2. Jabs, E.W., et al. 1994. Jackson-Weiss and Crouzon syndromes are allelic with mutations in fibroblast growth factor receptor 2. *Nat. Genet.* 8: 275-279.
3. Berger, P., et al. 2002. Molecular cell biology of Charcot-Marie-Tooth disease. *Neurogenetics* 4: 1-15.
4. Teresi, R.E., et al. 2007. Cowden syndrome-affected patients with PTEN promoter mutations demonstrate abnormal protein translation. *Am. J. Hum. Genet.* 81: 756-767.
5. Cho, M.Y., et al. 2008. First report of ovarian dysgerminoma in Cowden syndrome with germline PTEN mutation and PTEN-related 10q loss of tumor heterozygosity. *Am. J. Surg. Pathol.* 32: 1258-1264.
6. Yin, Y. and Shen, W.H. 2008. PTEN: a new guardian of the genome. *Oncogene* 27: 5443-5453.
7. Laugel, V., et al. 2010. Mutation update for the CSB/ERCC6 and CSA/ERCC8 genes involved in Cockayne syndrome. *Hum. Mutat.* 31: 113-126.

## CHROMOSOMAL LOCATION

Genetic locus: MCU (human) mapping to 10q22.1; Ccdc109a (mouse) mapping to 10 B4.

## SOURCE

CCDC109A (E-16) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of CCDC109A 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-246072 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

## APPLICATIONS

CCDC109A (E-16) is recommended for detection of CCDC109A of mouse, rat and 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); non cross-reactive with CCDC109B.

CCDC109A (E-16) is also recommended for detection of CCDC109A in additional species, including equine, canine, bovine, porcine and avian.

Suitable for use as control antibody for CCDC109A siRNA (h): sc-90666, CCDC109A siRNA (m): sc-142052, CCDC109A shRNA Plasmid (h): sc-90666-SH, CCDC109A shRNA Plasmid (m): sc-142052-SH, CCDC109A shRNA (h) Lentiviral Particles: sc-90666-V and CCDC109A shRNA (m) Lentiviral Particles: sc-142052-V.

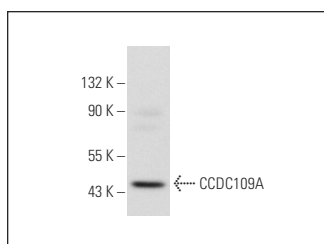
Molecular Weight of CCDC109A: 40 kDa.

Positive Controls: A549 cell lysate: sc-2413.

## 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



CCDC109A (E-16): sc-246072. Western blot analysis of CCDC109A expression in A549 whole cell lysate.

## RESEARCH USE

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