

# AMMECR1L (G-16): sc-169950

## BACKGROUND

AMMECR1 (Alport syndrome, mental retardation, midface hypoplasia and elliptocytosis chromosomal region gene 1), also known as AMME syndrome candidate gene 1 protein, is a 333 amino acid protein that contains one AMMECR1 domain. Encoded by a gene that maps to human chromosome Xq22.3, AMMECR1 is widely conserved, from *Caenorhabditis elegans* and yeast to microorganisms, with exon 2 encoding an evolutionarily conserved, six amino acid domain. Containing a glycine-rich N terminus, the AMMECR1 protein exhibits putative nuclear localization and a substantial level of instability, suggesting it plays a role in regulation. Additionally, numerous potential phosphorylation sites imply that AMMECR1 is subject to stringent regulation. AMMECR1 defects are linked to Alport syndrome, an X-linked contiguous gene deletion syndrome characterized by glomerulonephritis, deafness, mental retardation, midface hypoplasia and elliptocytosis.

## REFERENCES

- Piccini, M., et al. 1998. *FACL4*, a new gene encoding long-chain acyl-CoA synthetase 4, is deleted in a family with Alport syndrome, elliptocytosis, and mental retardation. *Genomics* 47: 350-358.
- Jonsson, J.J., et al. 1998. Alport syndrome, mental retardation, midface hypoplasia, and elliptocytosis: a new X linked contiguous gene deletion syndrome? *J. Med. Genet.* 35: 273-278.
- Vitelli, F., et al. 1999. Identification and characterization of a highly conserved protein absent in the Alport syndrome (A), mental retardation (M), midface hypoplasia (M), and elliptocytosis (E) contiguous gene deletion syndrome (AMME). *Genomics* 55: 335-340.
- Vitelli, F., et al. 2000. Identification and characterization of mouse orthologs of the AMMECR1 and *FACL4* genes deleted in AMME syndrome: orthology of Xq22.3 and MmuXF1-F3. *Cytogenet. Cell Genet.* 88: 259-263.
- Kashtan, C.E. 2000. Alport syndromes: phenotypic heterogeneity of progressive hereditary nephritis. *Pediatr. Nephrol.* 14: 502-512.
- Ferrante, M.I., et al. 2001. *IL1RAPL2* maps to Xq22 and is specifically expressed in the central nervous system. *Gene* 275: 217-221.

## CHROMOSOMAL LOCATION

Genetic locus: AMMECR1L (human) mapping to 2q14.3; *Ammecr1l* (mouse) mapping to 18 B1.

## SOURCE

AMMECR1L (G-16) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of AMMECR1L 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-169950 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

## APPLICATIONS

AMMECR1L (G-16) is recommended for detection of AMMECR1L 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).

AMMECR1L (G-16) is also recommended for detection of AMMECR1L in additional species, including equine, canine, bovine and avian.

Suitable for use as control antibody for AMMECR1L siRNA (h): sc-94373, AMMECR1L siRNA (m): sc-141050, AMMECR1L shRNA Plasmid (h): sc-94373-SH, AMMECR1L shRNA Plasmid (m): sc-141050-SH, AMMECR1L shRNA (h) Lentiviral Particles: sc-94373-V and AMMECR1L shRNA (m) Lentiviral Particles: sc-141050-V.

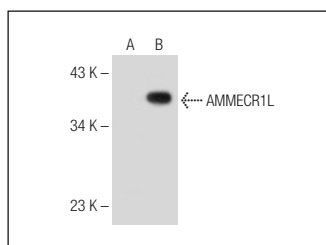
Molecular Weight of AMMECR1L: 36 kDa.

Positive Controls: AMMECR1L (m): 293T Lysate: sc-118379 or AMMECR1L (h): 293T Lysate: sc-117438.

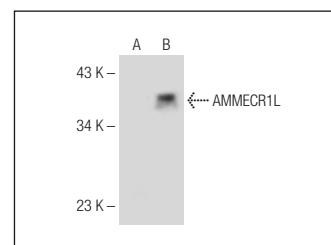
## RECOMMENDED SECONDARY REAGENTS

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



AMMECR1L (G-16): sc-169950. Western blot analysis of AMMECR1L expression in non-transfected: sc-117752 (A) and mouse AMMECR1L transfected: sc-118379 (B) 293T whole cell lysates.



AMMECR1L (G-16): sc-169950. Western blot analysis of AMMECR1L expression in non-transfected: sc-117752 (A) and human AMMECR1L transfected: sc-117438 (B) 293T whole cell lysates.

## 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.