SZABO SCANDIC

Part of Europa Biosite

Produktinformation

- Forschungsprodukte & Biochemikalien
- Zellkultur & Verbrauchsmaterial
- Diagnostik & molekulare Diagnostik
- Laborgeräte & Service

Weitere Information auf den folgenden Seiten!
See the following pages for more information!

Lieferung & Zahlungsart

siehe unsere Liefer- und Versandbedingungen

Zuschläge

- Mindermengenzuschlag
- Trockeneiszuschlag
- Gefahrgutzuschlag
- Expressversand

SZABO-SCANDIC HandelsgmbH

Quellenstraße 110, A-1100 Wien

T. +43(0)1 489 3961-0

F. +43(0)1 489 3961-7

mail@szabo-scandic.com

www.szabo-scandic.com

linkedin.com/company/szaboscandic

SANTA CRUZ BIOTECHNOLOGY, INC.

AMMECR1L (m): 293T Lysate: sc-118379

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

1. 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.
2. 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.
3. 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.
4. 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.
5. Kashtan, C.E. 2000. Alport syndromes: phenotypic heterogeneity of progressive hereditary nephritis. Pediatr. Nephrol. 14: 502-512.
6. Ferrante, M.I., et al. 2001. IL1RAPL2 maps to Xq22 and is specifically expressed in the central nervous system. Gene 275: 217-221.
7. Meloni, I., et al. 2002. Alport syndrome and mental retardation: clinical and genetic dissection of the contiguous gene deletion syndrome in Xq22.3 (ATS-MR). J. Med. Genet. 39: 359-365.
8. Tajika, Y., et al. 2005. Crystal structure of PH0010 from *Pyrococcus horikoshii,* which is highly homologous to human AMMECR 1C-terminal region. Proteins 58: 501-503.
9. Hertz, J.M. 2009. Alport syndrome. Molecular genetic aspects. Dan. Med. Bull. 56: 105-152.

CHROMOSOMAL LOCATION

Genetic locus: Ammecr1l (mouse) mapping to 18 B1.

PRODUCT

AMMECR1L (m): 293T Lysate represents a lysate of mouse AMMECR1L transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

APPLICATIONS

AMMECR1L (m): 293T Lysate is suitable as a Western Blotting positive control for mouse reactive AMMECR1L antibodies. Recommended use: 10-20 µl per lane.

Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

STORAGE

Store at -20° C. Repeated freezing and thawing should be minimized. Sample vial should be boiled once prior to use. 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 for detailed protocols and support products.