

# MLC1 (C-18): sc-86740

## BACKGROUND

MLC1 is a 377 amino acid multi-pass membrane protein that may serve as a non-selective neuronal cation channel in brain. Mutant MLC1 proteins that show impaired folding have been corrected *in vitro* with the addition of a Ca<sup>2+</sup>-ATPase inhibitor, curcumin. Mutations in the gene encoding MLC1 is the cause of megalencephalic leukoencephalopathy with subcortical cysts, also known as van der Knaap disease, a rare syndrome characterized early in life by progressive brain destruction causing mental retardation and inco-ordination. Single nucleotide polymorphisms within the MLC1 gene may be associated with periodic catatonia, but there seems to be conflicting evidence on whether or not the gene is implicated in general schizophrenia.

## REFERENCES

1. Leegwater, P.A., et al. 2001. Mutations of MLC1 (KIAA0027), encoding a putative membrane protein, cause megalencephalic leukoencephalopathy with subcortical cysts. *Am. J. Hum. Genet.* 68: 831-838.
2. Meyer, J., et al. 2001. A missense mutation in a novel gene encoding a putative cation channel is associated with catatonic schizophrenia in a large pedigree. *Mol. Psychiatry* 6: 302-306.
3. Leegwater, P.A., et al. 2002. Identification of novel mutations in MLC1 responsible for megalencephalic leukoencephalopathy with subcortical cysts. *Hum. Genet.* 110: 279-283.
4. Ben-Zeev, B., et al. 2002. Megalencephalic leukoencephalopathy with subcortical cysts; a founder effect in Israeli patients and a higher than expected carrier rate among Libyan Jews. *Hum. Genet.* 111: 214-218.
5. Rubie, C., et al. 2003. Sequence diversity of KIAA0027/MLC1: are megalencephalic leukoencephalopathy and schizophrenia allelic disorders? *Hum. Mutat.* 21: 45-52.
6. Gevaert, K., et al. 2003. Exploring proteomes and analyzing protein processing by mass spectrometric identification of sorted N-terminal peptides. *Nat. Biotechnol.* 21: 566-569.
7. Ilija Boor, P.K., et al. 2006. Megalencephalic leukoencephalopathy with subcortical cysts: an update and extended mutation analysis of MLC1. *Hum. Mutat.* 27: 505-512.
8. Selch, S., et al. 2007. MLC1 polymorphisms are specifically associated with periodic catatonia, a subgroup of chronic schizophrenia. *Biol. Psychiatry* 61: 1211-1214.

## CHROMOSOMAL LOCATION

Genetic locus: MLC1 (human) mapping to 22q13.33; Mlc1 (mouse) mapping to 15 E3.

## SOURCE

MLC1 (C-18) is an affinity purified rabbit polyclonal antibody raised against a peptide mapping near the C-terminus of MLC1 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 100 µg IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-86740 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

## APPLICATIONS

MLC1 (C-18) is recommended for detection of MLC1 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), immunohistochemistry (including paraffin-embedded sections) (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 MLC family members.

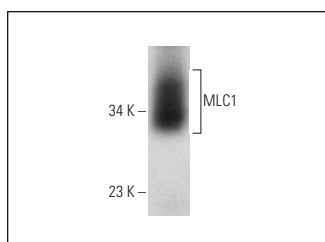
MLC1 (C-18) is also recommended for detection of MLC1 in additional species, including equine, canine and porcine.

Suitable for use as control antibody for MLC1 siRNA (h): sc-75793, MLC1 siRNA (m): sc-149463, MLC1 shRNA Plasmid (h): sc-75793-SH, MLC1 shRNA Plasmid (m): sc-149463-SH, MLC1 shRNA (h) Lentiviral Particles: sc-75793-V and MLC1 shRNA (m) Lentiviral Particles: sc-149463-V.

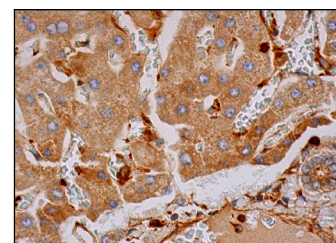
Molecular Weight of MLC1: 41 kDa.

Positive Controls: human brain hippocampus extract: sc-364375.

## DATA



MLC1 (C-18): sc-86740. Western blot analysis of MLC1 expression in human hippocampus tissue extract.



MLC1 (C-18): sc-86740. Immunoperoxidase staining of formalin fixed, paraffin-embedded human liver tissue showing cytoplasmic staining of hepatocytes, bile duct cells and hepatic sinusoids.

## SELECT PRODUCT CITATIONS

1. Tada, Y., et al. 2010. Characterization of cardiac size and function in SHRSPZ-Lep(fa)/IzmDmcr rats, a new animal model of metabolic syndrome. *Biol. Pharm. Bull.* 33: 1971-1976.

## 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.