



POMT1 (m): 293T Lysate: sc-127367

BACKGROUND

O-mannosylation is an essential protein modification in eukaryotes that is initiated by an evolutionarily conserved family of protein O-mannosyltransferases. The POMT1 (protein O-mannosyltransferase 1) protein consists of 725 amino acids. POMT1 contains 7 to 12 presumed transmembrane regions and a C-terminal ER membrane retention signal; RT-PCR reveals several mRNA splice variants. RNA dot blot analysis indicates ubiquitous expression of POMT1, with maximum levels in testis and high levels in fetal brain and pituitary tissues. Walker-Warburg syndrome (WWS), a severe, recessive, congenital muscular dystrophy associated with defects in neuronal migration that produce complex brain and eye abnormalities, is caused by mutations in the POMT1 gene.

REFERENCES

1. Akasaka-Manya, K., et al. 2004. Mutations of the POMT1 gene found in patients with Walker-Warburg syndrome lead to a defect of protein O-mannosylation. *Biochem. Biophys. Res. Commun.* 325: 75-79.
2. Ichimiya, T., et al. 2004. The twisted abdomen phenotype of *Drosophila* POMT1 and POMT2 mutants coincides with their heterophilic protein O-mannosyltransferase activity. *J. Biol. Chem.* 279: 42638-42647.
3. Willer, T., et al. 2004. Targeted disruption of the Walker-Warburg syndrome gene POMT1 in mouse results in embryonic lethality. *Proc. Natl. Acad. Sci. USA* 101: 14126-14131.
4. Yamamoto, T., et al. 2004. Expression and localization of fukutin, POMGnT1, and POMT1 in the central nervous system: consideration for functions of fukutin. *Med. Electron Microsc.* 37: 200-207.
5. Balci, B., et al. 2005. An autosomal recessive limb girdle muscular dystrophy (LGMD2) with mild mental retardation is allelic to Walker-Warburg syndrome (WWS) caused by a mutation in the POMT1 gene. *Neuromuscul. Disord.* 15: 271-275.
6. Currier, S.C., et al. 2005. Mutations in POMT1 are found in a minority of patients with Walker-Warburg syndrome. *Am. J. Med. Genet. A* 133: 53-57.
7. D'Amico, A., et al. 2006. Expanding the clinical spectrum of POMT1 phenotype. *Neurology* 66: 1564-1567.
8. Manya, H., et al. 2006. Molecular cloning and characterization of rat POMT1 and POMT2. *Glycobiology* 16: 863-873.
9. van Reeuwijk, J., et al. 2006. The expanding phenotype of POMT1 mutations: from Walker-Warburg syndrome to congenital muscular dystrophy, microcephaly and mental retardation. *Hum. Mutat.* 27: 453-459.

CHROMOSOMAL LOCATION

Genetic locus: Pomt1 (mouse) mapping to 2 B.

PRODUCT

POMT1 (m): 293T Lysate represents a lysate of mouse POMT1 transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

RESEARCH USE

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

APPLICATIONS

POMT1 (m): 293T Lysate is suitable as a Western Blotting positive control for mouse reactive POMT1 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.

PROTOCOLS

See our web site at www.scbt.com for detailed protocols and support products.